

Scientific and Technical Information Center

Requester's Full Name: NIRMA S. BASI Examiner #: 74538 Date: 3/27/03  
 Art Unit: 1666 Phone Number 30 89635 Serial Number: 09/768781  
 Mail Box and Bldg/Room Location: CML/10617 Results Format Preferred (circle): PAPER DISK E-MAIL

If more than one search is submitted, please prioritize searches in order of need. M.E.

Please provide a detailed statement of the search topic, and describe as specifically as possible the subject matter to be searched. Include the elected species or structures, keywords, synonyms, acronyms, and registry numbers, and combine with the concept or utility of the invention. Define any terms that may have a special meaning. Give examples or relevant citations, authors, etc, if known. Please attach a copy of the cover sheet, pertinent claims, and abstract.

Title of Invention: Isolated human Transport protein

Inventors (please provide full names): Gennady Merkulov

Earliest Priority Filing Date: 12/20/2000

\*For Sequence Searches Only\* Please include all pertinent information (parent, child, divisional, or issued patent numbers) along with the appropriate serial number.

- Please search
1. SEQID NO: 1, 2, and 3.
  2. nuclein acid molecule encoding the polypeptide of SEQID NO: 2.

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 commercial database  
 1-1350<sup>na</sup> issued patents  
 2-1389<sup>na</sup>  
 3-449<sup>aa</sup>

POINT OF CONTACT:  
 PAUL SCHULWITZ  
 TECHNICAL INFO. SPECIALIST  
 CM1 6B06 TEL. (703) 305-1954

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Searcher: \_\_\_\_\_  
 Searcher Phone #: \_\_\_\_\_  
 Searcher Location: \_\_\_\_\_  
 Date Searcher Picked Up: 3/28  
 Date Completed: 4/1  
 Searcher Prep & Review Time: 10  
 Clerical Prep Time: \_\_\_\_\_

Type of Search

NA Sequence (#) 1350 STN \_\_\_\_\_  
 AA Sequence (#) 1389 Dialog \_\_\_\_\_  
 Structure (#) 1320 Questel/Orbit \_\_\_\_\_  
 Bibliographic 1320 Dr. Link \_\_\_\_\_  
 Litigation \_\_\_\_\_ Lexis/Nexis \_\_\_\_\_  
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XKS.DWPI,TDBD,EPAB,JPAB,USPT,PGPB.	9
PROTEIN.DWPI,TDBD,EPAB,JPAB,USPT,PGPB.	273179
PROTEINS.DWPI,TDBD,EPAB,JPAB,USPT,PGPB.	169108
(XK ADJ PROTEIN).USPT,PGPB,JPAB,EPAB,DWPI,TDBD.	3
(XK PROTEIN).USPT,PGPB,JPAB,EPAB,DWPI,TDBD.	3

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<i>DB=USPT,PGPB,JPAB,EPAB,DWPI,TDBD; PLUR=YES; OP=ADJ</i>			
<u>L13</u>	xk protein	3	<u>L13</u>
<u>L12</u>	L2 and xk protein	1	<u>L12</u>
<u>L11</u>	L3 and xk protein	1	<u>L11</u>
<u>L10</u>	L9 and xk protein	1	<u>L10</u>
<u>L9</u>	beasley-ellen-m.in.	293	<u>L9</u>
<u>L8</u>	difranco-valentina.in.	0	<u>L8</u>
<u>L7</u>	di-franco-valentina.in.	0	<u>L7</u>
<u>L6</u>	di franco-valentina.in.	0	<u>L6</u>
<u>L5</u>	brandon-rhoda-c.in.	0	<u>L5</u>
<u>L4</u>	brandon-rhoda.in.	0	<u>L4</u>
<u>L3</u>	guegler-karl.in.	61	<u>L3</u>
<u>L2</u>	merkulov-gennady.in.	17	<u>L2</u>
<u>L1</u>	merkulov-geeady.in.	0	<u>L1</u>

END OF SEARCH HISTORY

FILE 'MEDLINE'  
FILE 'JAPIO'  
FILE 'BIOSIS'  
FILE 'SCISEARCH'  
FILE 'WPIDS'  
FILE 'CAPLUS'  
FILE 'EMBASE'  
=> s xk protein#  
L1 74 XK PROTEIN#

=> I1 and ligand transport  
L2 0 L1 AND LIGAND TRANSPORT

=> I1 and (transport# or transporter# or transporting)  
L3 29 L1 AND (TRANSPORT# OR TRANSPORTER# OR TRANSPORTING)

=> dup rem I3  
PROCESSING COMPLETED FOR L3  
L4 17 DUP REM L3 (12 DUPLICATES REMOVED)

=> dup rem I1  
PROCESSING COMPLETED FOR L1  
L5 36 DUP REM L1 (38 DUPLICATES REMOVED)

=> d I5 ibib abs 1-36

L5 ANSWER 1 OF 36 MEDLINE  
ACCESSION NUMBER: 2003138455 MEDLINE  
DOCUMENT NUMBER: 22540026 PubMed ID: 12652714  
TITLE: Cellulitis, sepsis, acute renal failure and hemolytic anemia with McLeod blood group phenotype.  
AUTHOR: Furiya Shino; Kitazawa Kunihiko; Ideura Gen; Toshida Fumitaka; Shimizu Shinsuke; Shimajo Takashi; Sakai Toshiaki; Ishiguro Jun; Miyahara Takashige; Misawa Takuo; Noguchi Osamu  
CORPORATE SOURCE: Department of Internal Medicine, Nagano Matsushiro General Hospital, Nagano.  
SOURCE: NIPPON NAIKA GAKKAI ZASSHI. JOURNAL OF JAPANESE SOCIETY OF INTERNAL MEDICINE, (2003 Jan 10) 92 (1) 140-2. Journal code: 19130210R. ISSN: 0021-5384.  
PUB. COUNTRY: Japan  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: Japanese  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200305  
ENTRY DATE: Entered STN: 20030326  
Last Updated on STN: 20030521  
Entered Medline: 20030520

L5 ANSWER 2 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.  
ACCESSION NUMBER: 2003:235497 BIOSIS  
DOCUMENT NUMBER: PREV200300235497  
TITLE: An attempt at analysing the selected traits of body conformation, growth, performance and genetic structure of Lithuanian native Zemaitukai horse, the breed being preserved from extinction.  
AUTHOR(S): Macijauskiene, Vale (1); Juras, Rytis (1)  
CORPORATE SOURCE: (1) Lithuanian Institute of Animal Science, R. Zebenkos 12, Baisogala, LT-5125, Radviliskio Raj., Lithuania Lithuania  
SOURCE: Animal Science Papers and Reports, (2003) Vol. 21, No. 1, pp. 35-46, print. ISSN: 0860-4037.  
DOCUMENT TYPE: Article  
LANGUAGE: English  
AB Over the last 50 years the traits of valuable indigenous Lithuanian Zemaitukai horse have not been investigated and during the last decade the breed became on the verge of extinction. Recently certain measures were undertaken to preserve the breed, evaluate its present characteristics and compare them with those reported earlier. Body size and conformation traits of present-day Zemaitukai horse (ZH) were found corresponding or similar to those of the ancient type, showing that many valuable characteristics of the breed are retained. Mares' milk yield and composition were evaluated, as well as development of foals based on body dimensions. Genetic variation, genetic structure as well as relationship between the lines and families of ZH were studied using blood typing and electrophoretic analysis of serum proteins. Gene frequencies are presented at six blood group (A, D, C, Q, P, K) and five protein (Al, Es, Gc, Xk, Tf) loci. The genetic diversity within blood groups and serum proteins in ZH kept in a closed population showed that out of eleven genetic systems examined, six were polymorphic. This is especially so for the A and D, as well as Es and Tf systems. The distribution of allele frequencies varied between the lines and families.

L5 ANSWER 3 OF 36 CAPLUS COPYRIGHT 2003 ACS  
ACCESSION NUMBER: 2002:755080 CAPLUS  
DOCUMENT NUMBER: 137:274161  
TITLE: Protein, gene and cDNA sequences of a novel human transport protein related to \*\*\*XK\*\*\*  
\*\*\*protein\*\*\* and their uses in drug screening  
INVENTOR(S): Merkulov, Gennady; Guegler, Karl; Brandon, Rhonda C.; Di Francesco, Valentina; Beasley, Ellen M.  
PATENT ASSIGNEE(S): USA  
SOURCE: U.S. Pat. Appl. Publ., 53 pp., Cont.-in-part of U.S. Ser. No. 740,034, abandoned. CODEN: USXXCO  
DOCUMENT TYPE: Patent  
LANGUAGE: English  
FAMILY ACC. NUM. COUNT: 2  
PATENT INFORMATION:

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
US 2002142376	A1	20021003	US 2001-768781	20010125
WO 2002072831	A2	20020919	WO 2002-US929	20020115

W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, OM, PH, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TN, TR, TT, TZ, UA, UG, UZ, VN, YU, ZA, ZM, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM  
RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG  
PRIORITY APPLN. INFO.: US 2000-740034 B2 20001220 US 2001-768781 A 20010125  
AB The invention provides protein, cDNA and genomic sequences for a novel human transport protein XK. The transport protein gene is expressed in human germinal center B cell. Eight single nucleotide polymorphism has been found on transport protein XK gene mapped to chromosome 23. The invention also relates to screening modulator of transport protein XK and use them in therapy. The invention further relates to methods, vector and hosts for expression of transport protein XK.

L5 ANSWER 4 OF 36 MEDLINE  
ACCESSION NUMBER: 2002480986 MEDLINE  
DOCUMENT NUMBER: 22228575 PubMed ID: 12243006  
TITLE: [Progress in molecular chorea diagnosis. McLeod syndrome and chorea acanthocytosis]. Fortschritte in der molekularen Chorea-Diagnostik. McLeod-Syndrom und Chorea-Akanthozytose.  
AUTHOR: Danek A  
CORPORATE SOURCE: Neurologische Klinik, Ludwig-Maximilians-Universitat, Postfach 701260, 81366 Munchen.. danek@nefro.med.uni-muenchen.de  
SOURCE: NERVENARZT, (2002 Jun) 73 (6) 564-9. Journal code: 0400773. ISSN: 0028-2804.  
PUB. COUNTRY: Germany: Germany, Federal Republic of  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: German  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200212  
ENTRY DATE: Entered STN: 20020924  
Last Updated on STN: 20021218  
Entered Medline: 20021217  
AB McLeod syndrome and chorea-acanthocytosis are classified with the so-called neuroacanthocytosis group of syndromes. Both lead to progressive basal ganglia degeneration and were not easily distinguished in the past. With the discovery of their molecular bases, mutations of the X-linked gene XK and autosomal recessive mutations of the gene coding for chorein, respectively, the two phenotypes can now be differentiated and extend the diagnostic spectrum in patients presenting with chorea. The present review compares the two conditions and proposes a practical approach to diagnosis and treatment. Better-defined disease concepts should eventually replace the umbrella term of "neuroacanthocytosis." Animal models are needed to understand the underlying mechanisms. A common pathway is likely for the pathogenesis of these conditions and is most probably shared with Huntington's disease.

L5 ANSWER 5 OF 36 CAPLUS COPYRIGHT 2003 ACS  
ACCESSION NUMBER: 2002:359541 CAPLUS  
DOCUMENT NUMBER: 137:214885  
TITLE: Point mutations causing the McLeod phenotype  
AUTHOR(S): Russo, David C. W.; Lee, Soohae; Reid, Marion E.;

Rodman, Colvin M.  
CORPORATE SOURCE: The New York Blood Center, Lindsley F. Kimball Research Institute, New York, NY, USA  
SOURCE: Transfusion (Malden, MA, United States) (2002), 42(3), 287-293  
CODEN: TRANAT; ISSN: 0041-1132  
PUBLISHER: Blackwell Publishing, Inc.  
DOCUMENT TYPE: Journal  
LANGUAGE: English  
AB The McLeod phenotype is defined by absence of Kx, weakening of Kell system antigens, and acanthocytosis. Individuals with the McLeod phenotype usually develop late-onset neuromuscular abnormalities. Gene deletions, insertions, and point mutations that affect RNA splicing or that lead to premature stop codons were reported to cause the McLeod phenotype. The McLeod phenotype may also be caused by mutations at a different splice site and by a novel mutation encoding an amino acid substitution that prevents transport to the cell surface. The coding and flanking intron regions of XK from 4 male, unrelated individuals with the McLeod phenotype and nonchronic granulomatous disease were sequenced and compared with the wild type sequence. Genomic DNA was amplified by PCR, and the products were sequenced. In 1 case, the mutant cDNA was expressed in a heterologous cell, and cell surface expression was deid. 3 Individuals with the McLeod phenotype had mutations that disrupted conserved GT sequences present at RNA splice sites. 2 Of them had G>C mutations at the 5' splice site of intron 1, and 1 had a G>A mutation at the 5' splice site of intron 2. One person with the McLeod phenotype had a 746C>G mutation in exon 3 encoding an R222G substitution. In a transfected cell, the expressed protein from the latter mutant did not travel to the cell surface. The McLeod phenotype may be caused by several different mutations.  
REFERENCE COUNT: 25 THERE ARE 25 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT  
L5 ANSWER 6 OF 36 EMBASE COPYRIGHT 2003 ELSEVIER SCI. B.V.  
ACCESSION NUMBER: 2002194888 EMBASE  
TITLE: [Differential diagnosis of hereditary chorea syndromes]. DIFFERENTIALDIAGNOSE HEREDITARER CHOREA-SYNDROME.  
AUTHOR: Jung H.H.  
CORPORATE SOURCE: Dr. H.H. Jung, Neurologische Klinik, Universitatsspital, Frauenklinikstrasse 26, CH-8091 Zurich, Germany. hans.jung@nos.uz.ch  
SOURCE: Schweizer Archiv fur Neurologie und Psychiatrie, (2002) 153/4 (185-188). Refs: 15 ISSN: 0258-7661 CODEN: SANPE8  
COUNTRY: Switzerland  
DOCUMENT TYPE: Journal; General Review  
FILE SEGMENT: 005 General Pathology and Pathological Anatomy  
008 Neurology and Neurosurgery  
022 Human Genetics  
032 Psychiatry  
LANGUAGE: German  
SUMMARY LANGUAGE: English  
AB The clinical triad of hereditary chorea syndromes includes (1) chorea/infant involuntary movement disorder, (2) psychiatric symptoms, and (3) cognitive impairment. The most frequent hereditary chorea syndrome is Huntington's disease (HD). There are several phenocopies of Huntington's disease, such as the Huntington's disease-like neurodegenerative disorders type 1 and type 2 (HDL2), benign hereditary chorea (BHC), dentato-rubro-pallido-Luysian atrophy (DRPLA), choreoacanthocytosis (CHAC), and McLeod syndrome (MLS). Huntington's disease is caused by an instable CAG trinucleotide expansion in the Huntington disease gene, and onset age and severity of symptoms depend on the number of CAG repeats. The physiological function of the gene product Huntingtin and the disease mechanisms are not fully elucidated yet. However, experimental data strongly suggest that induction of apoptosis through a caspase (cysteine aspartate-specific proteases)-dependent mechanism might be an important factor for the development of the striatal neurodegeneration. The HDL2s are more or less exact phenocopies of Huntington's disease. Two chromosomal localisations are described, and one responsible gene, Junctophilin-3, is identified. The BHC manifests as a pure chorea syndrome, without major psychiatric or cognitive impairment. The disease is located on chromosome 14, but the responsible gene has not yet been

identified. Apart from the Huntington's disease-like phenotype, DRPLA may manifest as a spinocerebellar ataxia, a progressive myoclonus epilepsy, or mixed phenotypes. DRPLA is caused by instable CAG expansions in Atrophin-1, whose physiological functions are not yet known. CHAC and MLS belong to the so-called neuroacanthocytosis syndromes. CHAC is an autosomal-recessive disorder characterised by a progressive chorea syndrome, perioral dyskinesias and mutilations, and - less frequently - an akinetic-rigid extrapyramidal syndrome and seizures. The responsible gene is located on chromosome 9, encoding chorein, a protein implicated in intracellular cell sorting. MLS is an X-linked multi-system disorder with haematological, neuromuscular, and CNS involvement. Haematologically, MLS is characterised by absent expression of the Kx erythrocyte antigen, weak expression of Kell antigens, acanthocytosis, and a compensated haemolytic state. Asymptomatic males have elevated serum creatine kinase levels, and are prone to develop neurological symptoms. Neuromuscular manifestations include myopathy, sensory-motor axonal neuropathy, and cardiomyopathy. CNS manifestations comprise a choreaiform movement disorder, neuropsychiatric abnormalities, and - less frequently - generalised seizures. MLS is caused by mutations of the XK gene encoding the \*\*\*XK\*\*\* protein\*\*\*, a putative membrane transport protein containing the Kx erythrocyte antigen. The \*\*\*XK\*\*\* protein\*\*\* is linked to the Kell glycoprotein by a single disulfide bond, probably forming a functional complex. The Kell protein is a member of the metalloproteinase family, and the \*\*\*XK\*\*\* protein\*\*\* has functional similarities to the CED-8 protein in nematodes, in which it controls the timing of apoptosis. These data strongly suggest an important role of the XK-Kell complex in striatal physiology. The advances in the molecular biology of hereditary chorea syndromes offer the possibility for a direct genetic analysis of affected individuals, and presymptomatic testing for individuals at risk. Although the genetic bases of some hereditary chorea syndromes are established, causal therapies are lacking. However, the rapidly accumulating knowledge will hopefully lead to the development of efficient therapies that might attenuate or even prevent these otherwise relentlessly progressive neurodegenerative disorders.

L5 ANSWER 7 OF 36 MEDLINE DUPLICATE 1  
 ACCESSION NUMBER: 2001423940 MEDLINE  
 DOCUMENT NUMBER: 21347986 PubMed ID: 11375401  
 TITLE: Molecular defects underlying the Kell null phenotype.  
 AUTHOR: Lee S; Russo D C; Reiner A P; Lee J H; Sy M Y; Telen M J;  
 Judd W J; Simon P; Rodrigues M J; Chabert T; Poole J;  
 Jovanovic-Szrenetic S; Levene C; Yahalom V; Redman C M  
 CORPORATE SOURCE: Lindsley F. Kimball Research Institute of the  
 New York Blood Center, New York, New York 10021, USA.  
 CONTRACT NUMBER: HL54459 (NHLBI)  
 SOURCE: JOURNAL OF BIOLOGICAL CHEMISTRY, (2001  
 Jul 20) 276 (29)  
 27281-9.  
 Journal code: 2985121R. ISSN: 0021-9258.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200108  
 ENTRY DATE: Entered STN: 20010827  
 Last Updated on STN: 20030105  
 Entered Medline: 20010823  
 AB Expression of the Kell blood group system is dependent on two proteins, Kell and XK, that are linked by a single disulfide bond. Kell, a type II membrane glycoprotein, is a zinc endopeptidase, while XK, which has 10 transmembrane domains, is a putative membrane transporter. A rare phenotype termed Kell null (Ko) is characterized by the absence of Kell protein and Kell antigens from the red cell membrane and diminished amounts of \*\*\*XK\*\*\* protein\*\*\*. We determined the molecular basis of eight unrelated persons with Ko phenotypes by sequencing the coding and the intron-exon splice regions of KEL and, in some cases, analysis of mRNA transcripts and expression of mutants on the cell surface of transfected cells. Six subjects were homozygous: four with premature stop codons, one with a 5' splice site mutation, G to A, in intron 3, and one with an amino acid substitution (S676N) in exon 18. Two Ko persons with premature stop codons had identical mutations in exon 4 (R128Stop), another had a different mutation in exon 4 (C83Stop), and the fourth had a stop codon in exon 9 (Q348Stop). Two Ko persons were heterozygous for two

mutations. One had a 5' splice site mutation (G to A) in intron 3 of one allele that caused aberrant splicing and exon skipping, and the other allele had an amino acid substitution in exon 10 (S363N). The other heterozygote had the same amino acid substitution in exon 10 (S363N) in one allele and a premature stop codon in exon 6 (R192Stop) in the other allele. The S363N and S676N mutants, expressed in 293T cells, were retained in a pre-Golgi compartment and were not transported to the cell surface, indicating that these mutations inhibit trafficking. We conclude that several different molecular defects cause the Kell null phenotype.

L5 ANSWER 8 OF 36 MEDLINE DUPLICATE 2  
 ACCESSION NUMBER: 2001514880 MEDLINE  
 DOCUMENT NUMBER: 21446863 PubMed ID: 11562915  
 TITLE: Kell and XK immunohistochemistry in McLeod myopathy.  
 AUTHOR: Jung H H; Russo D; Redman C; Brandner S  
 CORPORATE SOURCE: Department of Neurology, University Hospital Zurich, 8091 Zurich, Switzerland.. hans.jung@nos.usz.ch  
 CONTRACT NUMBER: HL54459 (NHLBI)  
 SOURCE: MUSCLE AND NERVE, (2001 Oct) 24 (10) 1346-51.  
 Journal code: 7803146. ISSN: 0148-639X.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200110  
 ENTRY DATE: Entered STN: 20010920  
 Last Updated on STN: 20021022  
 Entered Medline: 20011025  
 AB The McLeod syndrome is an X-linked neuroacanthocytosis manifesting with myopathy and progressive chorea. It is caused by mutations of the XK gene encoding the \*\*\*XK\*\*\* protein\*\*\*, a putative membrane transport protein of yet unknown function. In erythroid tissues, XK forms a functional complex with the Kell glycoprotein. Here, we present an immunohistochemical study in skeletal muscle of normal controls and a McLeod patient with a XK gene point mutation (C977T) using affinity-purified antibodies against XK and Kell proteins. Histological examination of the affected muscle revealed the typical pattern of McLeod myopathy including type 2 fiber atrophy. In control muscles, Kell immunohistochemistry stained sarcoplasmic membranes. XK immunohistochemistry resulted in a type 2 fiber-specific intracellular staining that was most probably confined to the sarcoplasmic reticulum. In contrast, there was only a weak background signal without a specific staining pattern for XK and Kell in the McLeod muscle. Our results demonstrate that the lack of physiological XK expression correlates to the type 2 fiber atrophy in McLeod myopathy, and suggest that the \*\*\*XK\*\*\* protein\*\*\* represents a crucial factor for the maintenance of normal muscle structure and function.  
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L5 ANSWER 9 OF 36 MEDLINE  
 ACCESSION NUMBER: 2001695801 MEDLINE  
 DOCUMENT NUMBER: 21612357 PubMed ID: 11746618  
 TITLE: The chorea of McLeod syndrome.  
 AUTHOR: Danek A; Tison F; Rubio J; Oechsner M; Kalkcreuth W; Monaco A P  
 CORPORATE SOURCE: Neurologische Klinik, Ludwig-Maximilians-Universitat, Munchen, Germany..  
 danek@brain.nfo.med.uni-muenchen.de  
 SOURCE: MOVEMENT DISORDERS, (2001 Sep) 16 (5) 882-9.  
 Journal code: 8610688. ISSN: 0885-3185.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200202  
 ENTRY DATE: Entered STN: 20011218  
 Last Updated on STN: 20021022  
 Entered Medline: 20020213  
 AB Among the movement disorders associated with acanthocytosis, McLeod syndrome (McKusick 314850) is the one that is best characterized on the molecular level. Its defining feature is low reactivity of Kell erythrocyte antigens. This is due to absence of membrane protein KX that forms a complex with the Kell protein. KX is coded for by the XK gene on the X-chromosome. We present six males (aged 29 to 60 years), with proven XK mutations, to discuss the chorea associated with McLeod syndrome. The movement disorder commonly develops in the fifth decade and is progressive. It affects the limbs, the trunk and the face. In addition to facial grimacing, involuntary vocalization can be present. In early stages there may only be some restlessness or slight involuntary distal movements of ankles and fingers. Lip-biting and facial tics seem more common in autosomal recessive choreoacanthocytosis linked to chromosome 9. This, together with the absence of dysphagia in McLeod syndrome, may

help in differential diagnosis. Recent findings suggest a role for the endothelin system of the striatum in the pathogenesis of McLeod syndrome.  
 Copyright 2001 Movement Disorder Society.

L5 ANSWER 10 OF 36 MEDLINE DUPLICATE 3  
 ACCESSION NUMBER: 2002035542 MEDLINE  
 DOCUMENT NUMBER: 21597008 PubMed ID: 11761473  
 TITLE: McLeod neuroacanthocytosis: genotype and phenotype.  
 AUTHOR: Danek A; Rubio J P; Rampoldi L; Ho M; Dobson-Stone C; Tison F; Symmans W A; Oechsner M; Kalkcreuth W; Watt J M; Corbett A J; Hamdalla H H; Marshall A G; Sutton I; Dotti M T; Malandrini A; Walker R H; Daniels G; Monaco A P  
 CORPORATE SOURCE: Neurologische Klinik, Ludwig-Maximilians-Universitat, Munchen, Germany..  
 danek@brain.nfo.med.uni-muenchen.de  
 SOURCE: ANNALS OF NEUROLOGY, (2001 Dec) 50 (6) 755-64.  
 Journal code: 7707449. ISSN: 0364-5134.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200201  
 ENTRY DATE: Entered STN: 20020124  
 Last Updated on STN: 20021022  
 Entered Medline: 20020107  
 AB McLeod syndrome is caused by mutations of XK, an X-chromosomal gene of unknown function. Originally defined as a peculiar Kell blood group variant, the disease affects multiple organs, including the nervous system, but is certainly underdiagnosed. We analyzed the mutations and clinical findings of 22 affected men, aged 27 to 72 years. Fifteen different XK mutations were found, nine of which were novel, including the one of the eponymous case McLeod. Their common result is predicted absence or truncation of the \*\*\*XK\*\*\* protein\*\*\*. All patients showed elevated levels of muscle creatine phosphokinase, but clinical myopathy was less common. A peripheral neuropathy with areflexia was found in all but 2 patients. The central nervous system was affected in 15 patients, as obvious from the occurrence of seizures, cognitive impairment, psychopathology, and choreatic movements. Neuroimaging emphasized the particular involvement of the basal ganglia, which was also detected in 1 asymptomatic young patient. Most features develop with age, mainly after the fourth decade. The resemblance of McLeod syndrome with Huntington's disease and with autosomal recessive chorea-acanthocytosis suggests that the corresponding proteins--XK, huntingtin, and chorein--might belong to a common pathway, the dysfunction of which causes degeneration of the basal ganglia.

L5 ANSWER 11 OF 36 MEDLINE DUPLICATE 4  
 ACCESSION NUMBER: 2001161085 MEDLINE  
 DOCUMENT NUMBER: 21157963 PubMed ID: 11261514  
 TITLE: McLeod syndrome: a novel mutation, predominant psychiatric manifestations, and distinct striatal imaging findings.  
 AUTHOR: Jung H H; Hergersberg M; Kneifel S; Alkadhi H; Schiess R; Weigell-Weber M; Daniels G; Kollias S; Hess K  
 CORPORATE SOURCE: Department of Neurology, University Hospital Zurich, Switzerland.. hans.jung@nos.usz.ch  
 SOURCE: ANNALS OF NEUROLOGY, (2001 Mar) 49 (3) 384-92.  
 Journal code: 7707449. ISSN: 0364-5134.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200104  
 ENTRY DATE: Entered STN: 20010425  
 Last Updated on STN: 20010425  
 Entered Medline: 20010419  
 AB The McLeod syndrome is an X-linked disorder caused by mutations of the XK gene encoding the \*\*\*XK\*\*\* protein\*\*\*. The syndrome is characterized by absent Kx erythrocyte antigen, weak expression of Kell blood group system antigens, and acanthocytosis. In some allelic variants, elevated creatine kinase, myopathy, neurogenic muscle atrophy, and progressive chorea are found. We describe a family with a novel point mutation in the XK gene consisting of a C to T base transition at nucleotide position 977, introducing a stop codon. Among seven affected males, five manifested with psychiatric disorders such as depression, bipolar disorder, or personality disorder, but only two presented with chorea Positron emission tomography and magnetic resonance volumetry revealed reduced striatal 2-fluoro-2-deoxy-glucose (FDG) uptake and

diminished volumes of the caudate nucleus and putamen that correlated with disease duration. In contrast, none of 12 female mutation carriers showed psychiatric or movement disorders. However, a semidominant effect of the mutation was suggested by erythrocyte and blood group mosaicism and reduced striatal FDG uptake without structural abnormalities. Therefore, patients with psychiatric signs or symptoms segregating in an X-linked trait should be examined for acanthocytosis and Kell/Kx blood group serology.

L5 ANSWER 12 OF 36 MEDLINE DUPLICATE 5  
ACCESSION NUMBER: 2001674195 MEDLINE  
DOCUMENT NUMBER: 21560274 PubMed ID: 11703337  
TITLE: A spontaneous novel XK gene mutation in a patient with McLeod syndrome.

AUTHOR: Supple S G; Iland H J; Barnett M H; Pollard J D  
CORPORATE SOURCE: The Kanematsu Laboratories, Royal Prince Alfred Hospital, Camperdown, NSW, Australia.

SOURCE: BRITISH JOURNAL OF HAEMATOLOGY, (2001 Nov) 115 (2) 369-72.

Journal code: 0372544. ISSN: 0007-1048.

PUB. COUNTRY: England; United Kingdom  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200112  
ENTRY DATE: Entered STN: 20011127  
Last Updated on STN: 20021022  
Entered Medline: 20011207

AB A 29-year-old man with a history of elevated creatine kinase and necrotizing myopathy was reviewed. Prominent red cell acanthocytosis in association with reduced Kell antigen expression was present, findings consistent with the McLeod syndrome. Investigation of the patient's XK gene revealed a novel TGG- to-TAG transition at position 1023 in exon 3.

This point mutation creates an in-frame stop codon (W314X), and predicts a truncated \*\*\*XK\*\*\* \*\*\*protein\*\*\* of 313 amino acids, compared with 444 amino acids in the normal \*\*\*XK\*\*\* \*\*\*protein\*\*\*. The mutation was not identified in the patient's mother or sister indicating that this mutation was spontaneous.

L5 ANSWER 13 OF 36 CAPLUS COPYRIGHT 2003 ACS  
ACCESSION NUMBER: 2001:934288 CAPLUS  
DOCUMENT NUMBER: 136:115930  
TITLE: Use of blood protein polymorphism for determining genetic distance between half-bred stallions  
AUTHOR(S): Pikula, Ryszard; Tomaszewska-Guszkiewicz, Krystyna;

Smugala, Mirosław; Gronet, Dominik  
CORPORATE SOURCE: Dep. of Horse Breeding, Agricultural Univ. of Szczecin, Szczecin, 71-466, Pol.  
SOURCE: Folia Universitatis Agriculturae Stetinensis (2001), 219, 67-71  
CODEN: FUASFI; ISSN: 1506-1965

PUBLISHER: Wydawnictwo Akademii Rolniczej w Szczecinie  
DOCUMENT TYPE: Journal  
LANGUAGE: English  
AB Genetic blood protein polymorphism of stallions was used to describe genetically 3 breeds of half-bred horses. The investigations covered Malopolski, Wielkopolski, and noble half-bred stallions from which blood samples were collected; in the samples, polymorphism of selected proteins: albumin (Al), transferrin (Tf), 8.5 pH esterase (EspH 8.5), vitamin D-binding protein (Gc), and \*\*\*Xk\*\*\* \*\*\*protein\*\*\* (Xk), was detd.

On the grounds of the performed studies, significant differences were found in phenotypic and allelic frequencies of blood protein systems analyzed according to the stallion breed. The av. heterozygosity and homozygosity coeffs. were established for stallion breeds as well as genetic similarity and genetic distance between breeds of the stallions. This distance was: 0.01046 between Malopolski and Wielkopolski stallions, 0.01783 between Malopolski and noble half-bred stallions, and 0.01000 between Wielkopolski and noble half-bred stallions.

REFERENCE COUNT: 7 THERE ARE 7 CITED REFERENCES  
AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L5 ANSWER 14 OF 36 MEDLINE DUPLICATE 6  
ACCESSION NUMBER: 2001063593 MEDLINE  
DOCUMENT NUMBER: 20553666 PubMed ID: 11099667  
TITLE: First example of anti-Kx in a person with the McLeod phenotype and without chronic granulomatous disease.  
AUTHOR: Russo D C; Oyen R; Powell V I; Perry S; Hitchcock J; Redman C M; Reid M E  
CORPORATE SOURCE: New York Blood Center, New York, New York, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: TRANSFUSION, (2000 Nov) 40 (11) 1371-5.  
Journal code: 0417360. ISSN: 0041-1132.

PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200012  
ENTRY DATE: Entered STN: 20010322  
Last Updated on STN: 20010322  
Entered Medline: 20001222  
AB BACKGROUND: Kx is lacking in the RBCs of patients with the McLeod syndrome. This condition is sometimes associated with chronic granulomatous disease (CGD). If given allogeneic RBCs, CGD patients with the McLeod phenotype may produce anti-Kx and anti-Km, and only phenotypically matched McLeod blood would be compatible. McLeod phenotype persons without CGD have made anti-Km but not anti-Kx (2 examples), and thus both McLeod and K(O) blood would be compatible. CASE REPORT: RBCs from a transfused patient with the McLeod phenotype but not with CGD (non-CGD McLeod) were typed for the Kell blood group antigens, and the plasma was analyzed for the presence of antibody by agglutination. The molecular basis was determined by analyzing for \*\*\*XK\*\*\* \*\*\*protein\*\*\* on RBC membranes by Western immunoblotting, by sequencing the XK gene, and by RFLP. RESULTS: The RBCs did not react with anti-Kx + anti-Km and showed weakening of Kell system antigens. The patient's plasma reacted moderately (2+) with RBCs of common Kell type and strongly (4+) with K(O) RBCs and RBCs of common Kell type treated with dithiothreitol, and did not react with McLeod RBCs. \*\*\*XK\*\*\* \*\*\*protein\*\*\* was absent from the RBC membranes. The XK gene had a point mutation in the donor splice site of intron 1 (G>C).  
CONCLUSION: This is the first report describing the molecular alteration in a non-CGD McLeod patient who has made anti-Kx. The immune response of people with the McLeod phenotype can vary, and K(O) blood may not always be compatible.

L5 ANSWER 15 OF 36 MEDLINE DUPLICATE 7  
ACCESSION NUMBER: 2000384103 MEDLINE  
DOCUMENT NUMBER: 20352021 PubMed ID: 10891471  
TITLE: Expression of Kell blood group protein in nonerythroid tissues.  
AUTHOR: Russo D; Wu X; Redman C M; Lee S  
CORPORATE SOURCE: Lindsley F. Kimball Research Institute, The New York Blood Center, New York, New York 10021, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: BLOOD, (2000 Jul 1) 96 (1) 340-6.  
Journal code: 7603509. ISSN: 0006-4971.

PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Abridged Index Medicus Journals; Priority Journals  
ENTRY MONTH: 200008  
ENTRY DATE: Entered STN: 20000818  
Last Updated on STN: 20021022  
Entered Medline: 20000810

AB The Kell blood group protein is a zinc endopeptidase that yields endothelin-3, a potent bioactive peptide, by cleavage of big endothelin-3, a larger intermediate precursor. On red cells, Kell protein is linked by a single disulfide bond to XK, a protein that traverses the membrane 10 times and whose absence, as occurs in the McLeod phenotype, is associated with a set of clinical symptoms that include nerve and muscle disorders and red cell acanthocytosis. Previous studies indicated that Kell is primarily expressed in erythroid tissues, whereas XK has a wider tissue distribution. The tissue distribution of Kell protein has been further investigated by Northern blot analysis, PCR-screening of tissue complementary DNAs (cDNAs), and Western immunoblots. Screening of an RNA dot-blot panel confirmed that Kell is primarily expressed in erythroid tissues but is also expressed in a near equal amount in testis, with weaker expression in a large number of other tissues. PCR-screening of cDNAs from different tissues and DNA sequencing of the products gave similar results. In 2 of the nonerythroid tissues tested, testis and skeletal muscle, Kell protein was detected by Western immunoblotting. In skeletal muscle, isolation of XK with a specific antibody coisolated Kell protein. These studies demonstrate that Kell is expressed in both erythroid and nonerythroid tissues and is associated with XK.

L5 ANSWER 16 OF 36 MEDLINE  
ACCESSION NUMBER: 2000384510 MEDLINE  
DOCUMENT NUMBER: 20307454 PubMed ID: 10849386  
TITLE: A murine monoclonal antibody against Kx protein which reacts also with beta-spectrin.  
AUTHOR: Carbonnet F; Blanchard D; Hattab C; Cochet S; Petit-Leroux Y; Loinat M J; Cartron J P; Bertrand O  
CORPORATE SOURCE: INSERM U76, Institut National de la Transfusion Sanguine, Alexandre Cabanel, Paris, France.

SOURCE: TRANSFUSION MEDICINE, (2000 Jun) 10 (2) 145-54.

Journal code: 9301182. ISSN: 0958-7578.  
PUB. COUNTRY: ENGLAND; United Kingdom  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200008  
ENTRY DATE: Entered STN: 20000818  
Last Updated on STN: 20021022  
Entered Medline: 20000808

AB Kx is a polytopic membrane protein of human erythrocytes carrying the Kx blood group antigen, which is deficient in rare patients with McLeod syndrome. Kx is disulphide bond linked to the Kell glycoprotein, which is a bitopic type II membrane protein carrying the Kell blood group antigen. Mice immunized with a synthetic peptide predicted to be located on the second external loop of Kx produced a monoclonal antibody called 3E12 which does not recognize red cells with common Kell phenotype by agglutination and flow cytometry. 3E12 recognizes the Kx protein and the spectrin beta-chain on western blots, the affinity for these two proteins being lowered with increasing ionic strength. Linear epitopes recognized by 3E12 are E116EIEKE121 and L484AQELEKE491 on the Kx protein and spectrin beta-chain, respectively. To quantify the relative amount of Kx in Empigen BB extracts of red cell membranes, an ELISA for Kx was set up which showed conclusively that (i) there is less Kx in membranes of K0 individuals (lacking the Kell glycoprotein) than in membranes of common individuals, and (ii) that all common individuals, typed as K+k-, K-k+ and K+k+, have the same amount of Kx on their red cell membranes. When an erythrocyte membrane detergent extract from one K0 individual was chromatographed on an immobilized 3E12 column, a minute amount of authentic Kell glycoprotein was recovered in acid eluted fractions, indicating that at least the K0 individual under study may still produce some Kell protein.

L5 ANSWER 17 OF 36 MEDLINE DUPLICATE 8  
ACCESSION NUMBER: 2000250542 MEDLINE  
DOCUMENT NUMBER: 20250542 PubMed ID: 10791880  
TITLE: The Kell blood group system: Kell and XK membrane proteins.  
AUTHOR: Lee S; Russo D; Redman C M  
CORPORATE SOURCE: Lindsley F. Kimball Research Institute, The New York Blood Center, New York 10021, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: SEMINARS IN HEMATOLOGY, (2000 Apr) 37 (2) 113-21. Ref: 62

Journal code: 0404514. ISSN: 0037-1963.  
PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
General Review; (REVIEW)  
(REVIEW, TUTORIAL)

LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200006  
ENTRY DATE: Entered STN: 20000629  
Last Updated on STN: 20021022  
Entered Medline: 20000621

AB Two membrane proteins express the antigens that comprise the Kell blood group system. A single antigen, Kx, is carried on XK, a 440-amino acid protein that spans the membrane 10 times, and more than 20 antigens reside on Kell, a 93-kd, type II glycoprotein. XK and Kell are linked, close to the membrane surface, by a single disulfide bond between Kell cysteine 72 and XK cysteine 347. Although primarily expressed in erythroid tissues, Kell and XK are also present in many other tissues. The polymorphic forms of Kell are due to single base mutations that encode different amino acids. Some Kell antigens are highly immunogenic and may cause strong reactions if mismatched blood is transfused and severe fetal anemia in sensitized mothers. Antibodies to KEL1 may suppress erythropoiesis at the progenitor level, leading to fetal anemia. The cellular functions of Kell/XK are complex. Absence of XK, the McLeod phenotype, is associated with acanthocytic red blood cells (RBCs), and with late-onset forms of muscular dystrophy and nerve abnormalities. Kell, by homology, is a member of the neprilysin (M13) family of membrane zinc endopeptidases and it preferentially activates endothelin-3 by specific cleavage of the Trp21-Ile22 bond of big endothelin-3.

L5 ANSWER 18 OF 36 MEDLINE DUPLICATE 9  
ACCESSION NUMBER: 2000244352 MEDLINE  
DOCUMENT NUMBER: 20244352 PubMed ID: 10782495  
TITLE: Functional and structural aspects of the Kell blood group system.  
AUTHOR: Lee S; Russo D; Redman C

CORPORATE SOURCE: Lindsley F Kimball Research Institute of the New York Blood Center, NY 10021, USA.

CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: TRANSFUSION MEDICINE REVIEWS, (2000 Apr) 14 (2) 93-103.

Ref: 49  
Journal code: 8709027. ISSN: 0887-7963.

PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
General Review; (REVIEW)  
(REVIEW, TUTORIAL)

LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200006  
ENTRY DATE: Entered STN: 20000629  
Last Updated on STN: 20021022  
Entered Medline: 20000616

AB Two covalently linked proteins, Kell and XK, constitute the Kell blood group system. Kell, a 93-Kd type II glycoprotein, is highly polymorphic and carries all but 1 of the known Kell antigens, and XK, which traverses the membrane 10 times, carries a single antigen, the ubiquitous Kx. The Kell/XK complex is not limited to erythroid tissues and may have multiple physiological roles. Absence of one of the component proteins, XK, is associated with abnormal red cell morphology and late-onset forms of nerve and muscle abnormalities, whereas the other protein component, Kell, is an enzyme whose principal known function is the production of a potent bioactive peptide, ET-3.

L5 ANSWER 19 OF 36 MEDLINE DUPLICATE 10  
ACCESSION NUMBER: 2001085975 MEDLINE  
DOCUMENT NUMBER: 21015021 PubMed ID: 11132157

TITLE: The mouse Kell blood group gene (Kel): cDNA sequence, genomic organization, expression, and enzymatic function.

AUTHOR: Lee S; Russo D C; Pu J; Ho M; Redman C M  
CORPORATE SOURCE: The Lindsley F. Kimball Research Institute of the New York

Blood Center, NY 10021, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: IMMUNOGENETICS, (2000 Nov) 52 (1-2) 53-62.  
Journal code: 0420404. ISSN: 0093-7711.

PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
OTHER SOURCE: GENBANK-AF252870  
ENTRY MONTH: 200101  
ENTRY DATE: Entered STN: 20010322  
Last Updated on STN: 20021022  
Entered Medline: 20010118

AB The human Kell blood group system is important in transfusion medicine, since Kell is a polymorphic protein and some of its antigens can cause severe reactions if mismatched blood is transfused, while maternal alloimmunization may lead to fetal and neonatal anemia. In humans, Kell is an Mr 93,000 type II membrane glycoprotein with endothelin-3-converting enzyme activity that is linked by a single disulfide bond to another protein, XK, that spans the membrane ten times. An absence of XK leads to clinical symptoms termed the McLeod syndrome. We determined the cDNA sequence of the mouse Kell homologue, the organization of the gene, expression of the protein and its enzymatic function on red cells. Comparison of human and mouse Kell cDNA showed 80% nucleotide and 74% amino acid sequence identity. Notable differences are that the mouse Kell protein has eight probable N-linked carbohydrate side chains, compared to five for human Kell, and that the mouse homologue has one more extracellular cysteine than human Kell protein. The mouse Kell gene (Kel), like its human counterpart, is similarly organized into 19 exons. Kel was located to proximal Chromosome 6. Northern blot analysis showed high expression in spleen and weaker levels in testis and heart. Western blot analysis of red cell membrane proteins demonstrated that mouse Kell glycoprotein has an apparent Mr of 110,000 and, on removal of N-linked sugars, 80,000. As in human red cells, Kell is disulfide-linked to XK and mouse red cells have endothelin-3-converting enzyme activity.

L5 ANSWER 20 OF 36 MEDLINE  
ACCESSION NUMBER: 2000411485 MEDLINE  
DOCUMENT NUMBER: 20353811 PubMed ID: 10895256  
TITLE: Kell, Kx and the McLeod syndrome.  
AUTHOR: Redman C M; Russo D; Lee S  
CORPORATE SOURCE: Laboratory of Membrane Biochemistry, Lindsley F. Kimball Research Institute, New York Blood Center, NY 10021, USA..  
credman@nybc.org  
SOURCE: Baillieres Best Pract Res Clin Haematol, (1999 Dec) 12 (4)

621-35. Ref: 95  
Journal code: 100900679. ISSN: 1521-6926.  
PUB. COUNTRY: ENGLAND: United Kingdom  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
General Review; (REVIEW)  
(REVIEW, TUTORIAL)

LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200008  
ENTRY DATE: Entered STN: 20000907  
Last Updated on STN: 20021022  
Entered Medline: 20000829

AB The antigens of the Kell blood group system are carried on a 93 kDa type II glycoprotein encoded by a single gene on chromosome 7 at 7q33. XK is a 50.9 kDa protein that traverses the membrane ten times and derives from a single gene on the X chromosome at Xp21. A single disulphide bond, Kell Cys 72-XK Cys 347, links Kell to XK. The Kell component of the Kell/XK complex is important in transfusion medicine since it is a highly polymorphic protein, carrying over 23 different antigens, that can cause severe reactions if mismatched blood is transfused and in pregnant mothers antibodies to Kell may elicit serious fetal and neonatal anaemia. The different Kell phenotypes are all caused by base mutations leading to single amino acid substitutions. By contrast the XK component carries a single blood group antigen, termed Kx. The physiological functions of Kell and XK have not been fully elucidated but Kell is a zinc endopeptidase with endothelin-3-converting enzyme activity and XK has the structural characteristics of a membrane transporter. Lack of Kx, the McLeod phenotype, is associated with red cell acanthocytosis, elevated levels of serum creatine phosphokinase and late onset forms of muscular and neurological defects.

L5 ANSWER 21 OF 36 MEDLINE  
ACCESSION NUMBER: 200009522 MEDLINE  
DOCUMENT NUMBER: 20009522 PubMed ID: 10541802  
TITLE: Structure and expression of the mouse homologue of the XK

gene.  
AUTHOR: Collec E; Colin Y; Carbonnet F; Hattab C; Bertrand O; Cartron J P; Kim C L  
CORPORATE SOURCE: INSERM U76, Institut National de la Transfusion Sanguine, 6 rue Alexandre Cabanel, 75015 Paris, France.  
SOURCE: IMMUNOGENETICS, (1999 Oct) 50 (1-2) 16-21.  
Journal code: 0420404. ISSN: 0093-7711.

PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
OTHER SOURCE: GENBANK-AF155511  
ENTRY MONTH: 199911  
ENTRY DATE: Entered STN: 20000111  
Last Updated on STN: 20021022  
Entered Medline: 19991119  
AB The human Kx blood group antigen is carried by a 37,000 M(r) apparent molecular mass membrane polypeptide which is deficient in rare individuals with the McLeod syndrome. The X-linked human XK gene is transcribed in many tissues including adult skeletal muscle and brain, sieges of disorders observed in McLeod syndrome. We report here the cloning of the orthologous mouse XK mRNA. Comparison of XK from human and mouse revealed 80% sequence similarity at the amino acid level. The mouse XK gene is organized in two exons and is expressed in many tissues, but its expression pattern is slightly different from that of the human gene. The presence in mouse erythrocyte membrane of a 43,000 M(r) Kx-related protein was demonstrated by immunoblotting with a rabbit antiserum directed against the human protein. With non-reduced samples, a 140,000 M(r) species was detected instead of the 43,000 M(r) protein, suggesting that, as demonstrated in the Kx polypeptide might be complexed with another protein in mouse red cells, presumably the homologue of the human Kell protein of 93,000 M(r).

L5 ANSWER 22 OF 36 MEDLINE DUPLICATE 11  
ACCESSION NUMBER: 2000025439 MEDLINE  
DOCUMENT NUMBER: 20025439 PubMed ID: 10556484  
TITLE: Intracellular assembly of Kell and XK blood group proteins.  
AUTHOR: Russo D; Lee S; Redman C  
CORPORATE SOURCE: Lindsley F. Kimball Research Institute, The New York Blood Center, 310 East 67 Street, New York, NY, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: BIOCHIMICA ET BIOPHYSICA ACTA, (1999 Nov 9) 1461 (1) 10-8.  
Journal code: 0217513. ISSN: 0006-3002.  
PUB. COUNTRY: Netherlands  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 199912  
ENTRY DATE: Entered STN: 20000113  
Last Updated on STN: 20021022  
Entered Medline: 19991227  
AB Kell, a 93 kDa type II membrane glycoprotein, and XK, a 444 amino acid multi-pass membrane protein, are blood group proteins that exist as a disulfide-bonded complex on human red cells. The mechanism of Kell/XK assembly was studied in transfected COS cells co-expressing Kell and \*\*\*XK\*\*\* \*\*\*proteins\*\*\*. Time course studies combined with endonuclease-H treatment and cell fractionation showed that Kell and XK are assembled in the endoplasmic reticulum. At later times the Kell component of the complex was not cleaved by endonuclease-H, indicating N-linked oligosaccharide processing and transport of the complex to a Golgi and/or a post-Golgi cell fraction. Surface-labeling of transfected COS cells, expressing both Kell and XK, demonstrated that the Kell/XK complex travels to the plasma membrane. XK expressed in the absence of Kell was also transported to the cell surface indicating that linkage of Kell and XK is not obligatory for cell surface expression.

L5 ANSWER 23 OF 36 SCISEARCH COPYRIGHT 2003 THOMSON ISI  
ACCESSION NUMBER: 1999:939262 SCISEARCH  
THE GENUINE ARTICLE: 260MH  
TITLE: Intracellular assembly of Kell and XK blood group proteins  
AUTHOR: Russo D; Lee S; Redman C (Reprint)  
CORPORATE SOURCE: NEW YORK BLOOD CTR, LINDSLEY F KIMBALL RES INST, 310 E 67 ST, NEW YORK, NY 10021 (Reprint); NEW YORK BLOOD CTR, LINDSLEY F KIMBALL RES INST, NEW YORK, NY 10021  
COUNTRY OF AUTHOR: USA  
SOURCE: BIOCHIMICA ET BIOPHYSICA ACTA-BIOMEMBRANES, (9 NOV 1999)  
Vol. 1461, No. 1, pp. 10-18.  
Publisher: ELSEVIER SCIENCE BV, PO BOX 211, 1000

AE AMSTERDAM, NETHERLANDS.  
ISSN: 0005-2736.  
DOCUMENT TYPE: Article; Journal  
FILE SEGMENT: LIFE  
LANGUAGE: English  
REFERENCE COUNT: 36  
\*ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS\*  
AB Kell, a 93 kDa type II membrane glycoprotein, and XK, a 444 amino acid multi-pass membrane protein, are blood group proteins that exist as a disulfide-bonded complex on human red cells. The mechanism of Kell/XK assembly was studied in transfected COS cells co-expressing Kell and \*\*\*XK\*\*\* \*\*\*proteins\*\*\*. Time course studies combined with endonuclease-H treatment and cell fractionation showed that Kell and XK are assembled in the endoplasmic reticulum. At later times the Kell component of the complex was not cleaved by endonuclease-H, indicating N-linked oligosaccharide processing and transport of the complex to a Golgi and/or a post-Golgi cell fraction. Surface-labeling of transfected COS cells, expressing both Kell and XK, demonstrated that the Kell/XK complex travels to the plasma membrane. XK expressed in the absence of Kell was also transported to the cell surface indicating that linkage of Kell and XK is not obligatory for cell surface expression. (C) 1999 Elsevier Science B.V. All rights reserved.

L5 ANSWER 24 OF 36 MEDLINE  
ACCESSION NUMBER: 1999353182 MEDLINE  
DOCUMENT NUMBER: 99353182 PubMed ID: 10426139  
TITLE: A novel frameshift mutation in the McLeod syndrome gene in a Japanese family.  
AUTHOR: Hanaoka N; Yoshida K; Nakamura A; Furihata K; Seo T; Tani Y; Takahashi J; Ikeda S; Hanyu N  
CORPORATE SOURCE: Department of Medicine (Neurology), Shinshu University School of Medicine, Matsumoto, Japan.  
SOURCE: JOURNAL OF THE NEUROLOGICAL SCIENCES, (1999 May 1) 165 (1) 6-9.  
Journal code: 0375403. ISSN: 0022-510X.

PUB. COUNTRY: Netherlands  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 199909  
ENTRY DATE: Entered STN: 19991005  
Last Updated on STN: 20021022  
Entered Medline: 19990920  
AB We report a novel mutation in the XK gene (XK) in a Japanese patient with McLeod syndrome. A 50-year-old man showed progressive muscular atrophy,

choreic movement, elevated level of serum creatinine kinase, and acanthocytosis. The expression level of all the Kell antigens in erythrocyte was decreased and molecular analysis revealed a single-base (T) deletion at the nucleotide position 1095 in XK. This deletion caused a frameshift in translation, leading to a premature stop codon at the amino acid position 408. We conclude this single-base deletion causes defective Kx protein, which is responsible for the McLeod phenotype in this patient.

L5 ANSWER 25 OF 36 MEDLINE DUPLICATE 12  
ACCESSION NUMBER: 1998256328 MEDLINE  
DOCUMENT NUMBER: 98256328 PubMed ID: 9593744  
TITLE: Association of XK and Kell blood group proteins.  
AUTHOR: Russo D; Redman C; Lee S  
CORPORATE SOURCE: Lindsley F. Kimball Research Institute, New York Blood Center, New York, New York 10021, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: JOURNAL OF BIOLOGICAL CHEMISTRY, (1998 May 29) 273 (22) 13950-6.  
Journal code: 2985121R. ISSN: 0021-9258.  
PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 199807  
ENTRY DATE: Entered STN: 19980713  
Last Updated on STN: 20021022  
Entered Medline: 19980701

AB A disulfide bond links Kell and XK red cell membrane proteins. Kell, a type II membrane glycoprotein, carries over 20 blood group antigens, and XK, which spans the membrane 10 times, is lacking in rare individuals with the McLeod syndrome. Kell is classified in the neprilysin family of zinc endopeptidases, and XK has structural features that suggest it is a transport protein. Kell has 15 extracellular cysteines, and XK has one in

its fifth extracellular loop. Five of the extracellular cysteine residues in Kell are not conserved in the other members of the neprilysin family, and based on the hypothesis that one of the nonconserved cysteines is linked to XK, cysteines 72 and 319 were mutated to serine. The single extracellular cysteine 347 of XK was also mutated. Co-expression of combinations of wild-type and mutant proteins in transfected COS-1 cells showed that Kell C72S did not form a Kell-XK complex with wild-type XK, while wild-type Kell and Kell C319S did. XK C347S was also unable to form a complex with wild-type Kell, indicating that Kell cysteine 72 is linked to XK cysteine 347. Kell C72S was transported to the cell surface, indicating that linkage to XK is not required. In addition, chemical cross-linking of red cell membranes with dithiobispropionimide indicated that glyceraldehyde-3-phosphate dehydrogenase is a near neighbor of Kell.

L5 ANSWER 26 OF 36 MEDLINE  
ACCESSION NUMBER: 1999003496 MEDLINE  
DOCUMENT NUMBER: 99003496 PubMed ID: 9784384  
TITLE: Kx, a quantitatively minor protein from human erythrocytes, is palmitoylated in vivo.  
AUTHOR: Carbonnet F; Hattab C; Callebaut I; Cochet S; Blancher A; Cartron J P; Bertrand O  
CORPORATE SOURCE: Institut National de la Transfusion Sanguine, 6 rue Alexandre Cabanel, Paris, 75015, France.  
SOURCE: BIOCHEMICAL AND BIOPHYSICAL RESEARCH COMMUNICATIONS, (1998 Sep 29) 250 (3) 569-74.  
Journal code: 0372516. ISSN: 0006-291X.  
PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 199811  
ENTRY DATE: Entered STN: 19990106  
Last Updated on STN: 20021022  
Entered Medline: 19981123

AB Kx is a quantitatively minor blood group protein of human erythrocytes which is thought to be a membrane transporter. In the red cell membrane, Kx forms a complex stabilized by a disulfide bond with the Kell blood group membrane protein which might function as a metalloprotease. The palmitoylation status of these proteins was studied by incubating red cells with [3H] palmitic acid. Purification of the Kell-Kx complex, by immunochromatography on an immobilized human monoclonal antibody of Kell blood group specificity demonstrated that the Kx but not the Kell protein is palmitoylated. Six cysteines in Kx are predicted to be intracytoplasmic and might be targets for palmitoylation. Three of these cysteines are present in a portion of sequence which is predicted to form an amphipathic alpha helix. Palmitoylation of one or several of these cysteines might contribute to anchor the cytoplasmic portion of the Kx

protein to the inner surface of red cell membrane.  
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L5 ANSWER 27 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.  
ACCESSION NUMBER: 2000:267961 BIOSIS  
DOCUMENT NUMBER: PREV200000267961  
TITLE: Genetic analysis in the Basque Pony-Pottoka breed: Preliminary results.  
AUTHOR(S): Pascual Moro, I. (1); Tejedor, T.; Monteagudo Ibanez, L. V.; Intxausti del Casal, J. I. (1); Arruga Lavina, M. V.  
CORPORATE SOURCE: (1) Servicio de Ganaderia, Diputacion Foral de Bizkaia, Lehendakari Agirre Etorbidea, 9, 2, 48014, Bilbao Spain  
SOURCE: Archivos de Zootecnia, (1998) Vol. 47, No. 178-179, pp. 181-188. print.. Meeting Info.: Spanish Society for the animal Genetic Resources. Cordoba, Spain December 14-17, 1997 Nacional de la Sociedad Espanola para los Recursos Geneticos Animales ISSN: 0004-0592.  
DOCUMENT TYPE: Conference  
LANGUAGE: Spanish  
SUMMARY LANGUAGE: English

L5 ANSWER 28 OF 36 SCISEARCH COPYRIGHT 2003 THOMSON ISI  
ACCESSION NUMBER: 97:609377 SCISEARCH  
THE GENUINE ARTICLE: XQ325  
TITLE: Analysis of the McLeod syndrome gene in three patients with neuroacanthocytosis  
AUTHOR: Shizuka M; Watanabe M; Aoki M; Ikeda Y; Mizushima K; Okamoto K; Itoyama Y; Abe K; Shoji M (Reprint)  
CORPORATE SOURCE: GUNMA UNIV, SCH MED, DEPT NEUROL, 3-39-15 SHOWA MACHI, MAEBASHI, GUMMA 371, JAPAN (Reprint); GUNMA UNIV, SCH MED, DEPT NEUROL, MAEBASHI, GUMMA 371, JAPAN; TOHOKU UNIV, SCH MED, DEPT NEUROL, SENDAI, MIYAGI 980, JAPAN  
COUNTRY OF AUTHOR: JAPAN  
SOURCE: JOURNAL OF THE NEUROLOGICAL SCIENCES, (10 SEP 1997) Vol. 150, No. 2, pp. 133-135.  
Publisher: ELSEVIER SCIENCE BV, PO BOX 211, 1000 AMSTERDAM, NETHERLANDS.  
ISSN: 0022-510X.  
DOCUMENT TYPE: Article; Journal  
FILE SEGMENT: LIFE  
LANGUAGE: English  
REFERENCE COUNT: 8  
\*ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS\*

AB McLeod syndrome is a rare X-linked disorder involving neurological defects and acanthocytosis. We examined the XK gene in three patients with neuroacanthocytosis, one of whom had cardiomyopathy, and his symptoms were very similar to those of McLeod syndrome. We found two new transversions (C to G at codon 204 and G to C at codon 205) in exon 3 in all those cases. However, the transversion at codon 205 was found in all 70 Japanese normal subjects and four non-Japanese (two Caucasian males, one Chinese female and one Micronesian female) and that at codon 204 was also detected in all 14 normal Japanese males and the four non-Japanese. These findings suggest that they are not the cause of McLeod syndrome, but normal polymorphisms which have not been reported. Moreover, there is a possibility that patients with neuroacanthocytosis similar to McLeod syndrome exist without the XK gene abnormalities. (C) 1997 Elsevier Science B.V.

L5 ANSWER 29 OF 36 MEDLINE DUPLICATE 13  
ACCESSION NUMBER: 94273191 MEDLINE  
DOCUMENT NUMBER: 94273191 PubMed ID: 8004674  
TITLE: Isolation of the gene for McLeod syndrome that encodes a novel membrane transport protein.  
AUTHOR: Ho M; Chelly J; Carter N; Danek A; Crocker P; Monaco A P  
CORPORATE SOURCE: Imperial Cancer Research Fund Laboratories, Institute of Molecular Medicine, John Radcliffe Hospital, Oxford, England.  
SOURCE: CELL, (1994 Jun 17) 77 (6) 869-80.  
Journal code: 0413066. ISSN: 0092-8674.  
PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
OTHER SOURCE: GENBANK-Z32684  
ENTRY MONTH: 199407  
ENTRY DATE: Entered STN: 19940729  
Last Updated on STN: 20021022  
Entered Medline: 19940721

AB McLeod syndrome is an X-linked multisystem disorder characterized by abnormalities in the neuromuscular and hematopoietic systems. We have assembled a cosmid contig of 360 kb that encompasses the McLeod gene locus. A 50 kb deletion was detected by screening DNA from patients with radiolabeled whole cosmids, and two transcription units were identified within this deletion. The mRNA expression pattern of one of them, designated as XK, correlates closely to the McLeod phenotype. XK encodes a novel protein with structural characteristics of prokaryotic and eukaryotic membrane transport proteins. Nucleotide sequence analysis of XK from two unrelated McLeod patients has identified point mutations at conserved splice donor and acceptor sites. These findings provide direct evidence that XK is responsible for McLeod syndrome.

L5 ANSWER 30 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 14  
ACCESSION NUMBER: 1995:58381 BIOSIS  
DOCUMENT NUMBER: PREV199598072681  
TITLE: Efficiency of some serum protein systems in parentage control in Yugoslav trotter horses.  
AUTHOR(S): Trailovic, Ruzica; Jovanovic, S.; Savic, Mila  
CORPORATE SOURCE: Fac. Vet. Med., Univ. Belgrade, Bul. JNA 18, Belgrade Yugoslavia  
SOURCE: Acta Veterinaria (Belgrade), (1994) Vol. 44, No. 4, pp. 233-237.  
ISSN: 0567-8315.

DOCUMENT TYPE: Article  
LANGUAGE: English  
SUMMARY LANGUAGE: English; Serbo-Croatian  
AB A total of 85 blood samples, obtained from Yugoslav trotter horses Were analysed for serum protein polymorphism at the following loci: albumin (Al), protease inhibitor (Pi), transferrin (Tf), esterase (Es) and \*\*\*Xk\*\*\* \*\*protein\*\*\* by standard starch gel electrophoretic procedures. From the results obtained the homogeneity index and parentage exclusion probability were calculated. The characteristic gene frequencies of the investigated Al, Pi, Ti, Es and \*\*\*Xk\*\*\* \*\*protein\*\*\* systems were established as: AIA and AIB (0.424 and 0.576); PiF, PiL, PiG, PiH, PiV and PiS (0.135, 0.318, 0.123, 0.100, 0.259 and 0.576); TiD, TiF, TiH and TiO (0.359, 0.529, 0.036 and 0.076), EsF, EsI and EsS (0.265, 0.570 and 0.165); and XkK and XkS (0.912 and 0.088), respectively. The Homogeneity index of the tested population was 0.0049, 0.5755, 0.2209, 0.1336 and 0.6790 for the AL, Pi, Tf, Es and Xk, loci, respectively. The joint paternity exclusion probability was 83.40% for the population of Yugoslav trotters.

L5 ANSWER 31 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 15  
ACCESSION NUMBER: 1993:294126 BIOSIS  
DOCUMENT NUMBER: PREV199396012351  
TITLE: A study on the polymorphism of blood protein and enzyme in Cheju native horses.  
AUTHOR(S): Kim, Sang-Yong; Kang, Min-Soo; Choung, Chang-Cho; Takahashi, Jutaro; Yasuda, Yasuhisa  
CORPORATE SOURCE: Fac. Agric. Iwate Univ., Morioka Japan  
SOURCE: Journal of the Faculty of Agriculture Iwate University, (1993) Vol. 21, No. 2, pp. 91-96.  
ISSN: 0579-2746.

DOCUMENT TYPE: Article  
LANGUAGE: Japanese  
SUMMARY LANGUAGE: Japanese; English  
AB On the basis of gene frequencies of the marker traits of blood protein and enzyme analyses with electrophoresis, the biochemical polymorphism of albumin (Al), slow-alpha-2 globulin (S1-alpha), post-albumin (Pa), group-specific component (Gc), \*\*\*Xk\*\*\* \*\*protein\*\*\* (Xk), transferrin (Tf), catalase (Cat), hemoglobin (Hb), phosphohexose isomerase (PHI), phosphoglucuronate dehydrogenase (PGD) and phosphoglucosutase (PGM), in a total 95 Cheju native horses, were examined. The analyzed results of phenotypes and gene frequencies were as follows: With respect to albumin (Al) locus, the frequency of Al-B allele was markedly predominant (0.663) as compared with that of Al-A allele (0.337). In slow alpha-2 globulin (S1-alpha-2) locus, any individual variation was not found. Therefore, this locus was defined to be monomorphic. In the post-albumin (Pa) locus, the frequency of Pa-F allele was markedly predominant (0.947) as compared with that of Pa-S allele (0.053). Concerning group-specific component (Gc)

locus, the frequency of Gc-S allele was markedly predominant (0.589) as compared with that of Gc-F allele (0.441). As to the \*\*\*Xk\*\*\* protein\*\*\* locus, one phenotype KK was observed. The number of the KK phenotype was 1.000. In the transferrin (Tf) locus, Tf-F was the most frequent allele gene frequency (0.621), Tf-R was the second (0.153) and Tf-H, Tf-D and Tf-O were negligible (0.131, 0.084, and 0.010). At the catalase (Cat) isozyme locus, the gene frequency of Cat-F allele (0.511) was slightly higher than that of Cat-S allele (0.489). In the hemoglobin (Hb) locus, the frequency of Hb-A allele (0.868) was remarkably higher than that of Hb-a allele (0.132). At the phosphohexose isomerase (PHI) isozyme locus, only phenotype II was observed. The frequency of the II type was 1.000. Phosphoglucomutase (PGM) isozyme locus, any individual variation was not found. As to phosphogluconate dehydrogenase (PGD) isozyme locus, any individual variation was not found.

L5 ANSWER 32 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 16  
 ACCESSION NUMBER: 1990:471454 BIOSIS  
 DOCUMENT NUMBER: BA90:110874  
 TITLE: STUDIES ON THE BIOCHEMICAL POLYMORPHISM OF BLOOD PROTEIN AND ENZYME IN CHE JU NATIVE HORSES I. GENETIC POLYMORPHISMS OF SERUM PROTEINS.  
 AUTHOR(S): CHUNG E Y; HAN S K; SHIN Y C; YANG K S  
 CORPORATE SOURCE: COLL. AGRIC., SANG JI UNIV., KOREAN.  
 SOURCE: KOREAN J ANIM SCI, (1990) 32 (6), 298-308.  
 CODEN: HGCHAG. ISSN: 0367-5807.  
 FILE SEGMENT: BA; OLD  
 LANGUAGE: Korean  
 AB By means of starch gel electrophoresis, the biochemical polymorphism of .alpha.1-protease inhibitor, albumin, transferrin, \*\*\*Xk\*\*\* protein\*\*\* and slow .alpha.2-globulin in a total of 116 Che Ju native horses were examined. The analyzed resulted of phenotype, genotype and gene frequency was following: 1. In the .alpha.1-protease inhibitor(Pi) locus, nine possible phenotypes, except heterozygous F1 phenotype, FF, LL, SS, FL, FS, IL, IS and LS were identified and assumed to be controlled by four autosomal codominant alleles designated PiF, PiL, PiI and PiS. The phenotype distribution was estimated to be 68.10% for LL type and 12.93% for II type and the others were below 10%. The PiL allele with the frequency of 0.741 showed the highest frequency, while the frequencies of PiI, PiS and PiF alleles with relatively low frequencies were 0.164, 0.078 and 0.017, respectively. 2. With respect to albumin(AI) locus, three different AI phenotypes assumed to be controlled by two codominant alleles were identified as AA, AB and BB and their phenotype distribution was 15.52%, 40.52% and 43.96%, respectively. The frequency of AIB allele was markedly predominant (0.641) whereas in A1A allele it was 0.358. 3. Concerning transferrin(Tf) locus, eleven different phenotypes DD, FF, RR, DF, DO, DR, FH, FO, FR, HR and OR were recognized, assumed to be controlled by five autosomal codominant alleles designated TFD, TfF, TfH, TfO and TfR, but two homozygous type(HH and OO) and two heterozygous type(DH and HO) were not found. The observed percentage of Tf phenotypes FR, FF and RR were found to be 29.31%, 28.45% and 12.93%, respectively, and the other phenotypes were below 10%. Of the total, TfF was the most frequent allele(gene frequency, 0.496), TfR was the second(0.345) and TFD, TfO and TfH were negligible(0.065, 0.60 and 0.034, respectively). 4. As for the \*\*\*Xk\*\*\* protein\*\*\* locus, two different phenotypes FK and KK were observed, whereas homozygous FF type was not recognized. The observed Xk polymorphism was assumed to be controlled by a pair of codominant alleles designated XkF and XkK at a single autosomal locus. The number of the KK phenotype was 93.10, that of FK phenotype 6.90%. The significantly higher frequency of XkK allele(0.966) was obtained than that of XkF allele(0.034). 5. In slow .alpha.2-globulin(S .alpha.1) locus, any individual variation was not found, therefore, this locus was defined to be monomorphic.

L5 ANSWER 33 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.  
 ACCESSION NUMBER: 1990:284226 BIOSIS  
 DOCUMENT NUMBER: BA90:15072  
 TITLE: STUDIES ON BLOOD GROUPS IN RACE HORSES V. GENETIC POLYMORPHISM OF SERUM \*\*\*XK\*\*\* PROTEIN\*\*\* .

AUTHOR(S): HAN S K; CHUNG E Y; KANG H I  
 CORPORATE SOURCE: COLL. ANIMAL HUSBANDRY, KON-KUK UNIV., JPN.  
 SOURCE: KOREAN J ANIM SCI, (1990) 32 (2), 61-65.  
 CODEN: HGCHAG. ISSN: 0367-5807.  
 FILE SEGMENT: BA; OLD  
 LANGUAGE: Japanese  
 AB Genetic polymorphism of a new horse plasma protein provisionally designated \*\*\*Xk\*\*\* protein\*\*\* in 175 Korean race horses was analyzed by using acidic starch gel electrophoresis and genetic structure of horse population was investigated. Two different phenotypes, Xk-FK and Xk-KK. in this system were observed with the frequencies in these Xk phenotypes were Xk-FK 2.9% and Xk-KK 97.1%. However, the homozygous Xk-FF type was not recognized in the present study. Observed and expected phenotypes showed the Xk locus to be in genetic equilibrium, according to Hardy-Weinberg law. Therefore, the Xk phenotypes were shown to be controlled by two codominant autosomal alleles designated XkF and XkK. The XkK allele(0.986) had a remarkably high frequency whereas the XkF allele(0.014) occur very rarely.

L5 ANSWER 34 OF 36 MEDLINE DUPLICATE 17  
 ACCESSION NUMBER: 89250430 MEDLINE  
 DOCUMENT NUMBER: 89250430 PubMed ID: 3248368  
 TITLE: The homology between the serum proteins PO2 in pig, Xk in horse and alpha 1B-glycoprotein in human.  
 AUTHOR: Van de Weghe A; Coppieters W; Bauw G; Vandekerckhove J; Bouquet Y  
 CORPORATE SOURCE: Department of Animal Genetics, State University of Ghent, Merelbeke, Belgium.  
 SOURCE: COMPARATIVE BIOCHEMISTRY AND PHYSIOLOGY. B: COMPARATIVE BIOCHEMISTRY, (1988) 90 (4) 751-6.  
 Journal code: 2984730R. ISSN: 0305-0491.  
 PUB. COUNTRY: ENGLAND: United Kingdom  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 198906  
 ENTRY DATE: Entered STN: 19900306  
 Last Updated on STN: 19900306  
 Entered Medline: 19890628  
 AB 1. Pig serum Po2 protein and horse \*\*\*Xk\*\*\* protein\*\*\* were purified by FPLC, non-denaturing 2D agarose-PAGE and 2D IPG-PAGE. 2. The separated fractions were electroblotted to poly(4-vinyl-N-methylpyridinium iodide) coated GF/C glass fiber sheets. 3. The partial amino acid sequences and amino acid compositions of different genetic variants of the proteins were determined. 4. The results proved that previously reported polymorphic serum post-albumins in each of these species were homologous to human plasma alpha 1B-glycoprotein.

L5 ANSWER 35 OF 36 MEDLINE DUPLICATE 18  
 ACCESSION NUMBER: 83306728 MEDLINE  
 DOCUMENT NUMBER: 83306728 PubMed ID: 6614593  
 TITLE: Genetic linkage between the loci for phosphohexose isomerase (PHI) and a serum protein (Xk) in horses.  
 AUTHOR: Andersson L; Juneja R K; Sandberg K  
 SOURCE: ANIMAL BLOOD GROUPS AND BIOCHEMICAL GENETICS, (1983) 14 (1) 45-50.  
 Journal code: 0263344. ISSN: 0003-3480.  
 PUB. COUNTRY: Netherlands  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 198310  
 ENTRY DATE: Entered STN: 19900319  
 Last Updated on STN: 19980206  
 Entered Medline: 19831028  
 AB Genetic linkage between the equine loci for phosphohexose isomerase (PHI) and serum \*\*\*Xk\*\*\* protein\*\*\* was demonstrated by means of segregation data from three sire families. The recombination frequency was estimated from pooled data to be 0.23 +/- 0.02; a significant heterogeneity between sires for estimates of the recombination frequency was observed. No indication of linkage was detected between Xk and 14 other blood marker loci. Linkage between the Xk locus and the locus for soluble malic enzyme (ME1) has recently been reported in horses. An equine linkage group designated LG IV comprising the three loci ME1, PHI, and Xk has thus been established. The possibility that the linkage between PHI and Xk is homologous to the linkage between the loci for PHI and a serum postalbumin (PO-2) in pigs was discussed.

L5 ANSWER 36 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.  
 ACCESSION NUMBER: 1984:185837 BIOSIS  
 DOCUMENT NUMBER: BA77:18821  
 TITLE: EQUINE GENE MAPPING CLOSE LINKAGE BETWEEN THE LOCI FOR SOLUBLE MALIC ENZYME EC-1.1.1.40 AND XK PA.  
 AUTHOR(S): WEITKAMP L R; COSTELLO-LEARY P; GUTTORMSEN S A  
 CORPORATE SOURCE: DEP. PSYCHIATRY, DIV. GENETICS, UNIV. ROCHESTER SCH. MED. DENT., 601 ELMWOOD AVE., ROCHESTER, N.Y. 14642, U.S.A.  
 SOURCE: ANIM BLOOD GROUPS BIOCHEM GENET, (1982 (RECD 1983)) 13 (4), 279-284.  
 CODEN: ABBGBX. ISSN: 0003-3480.  
 FILE SEGMENT: BA; OLD  
 LANGUAGE: English  
 AB Resolution of equine soluble malic enzyme phenotypes is greatly improved by isoelectric focusing as compared with starch gel electrophoresis. Phenotype differences can be recognized in plasma as well as hemolysates. The locus for soluble malic enzyme (ME1) is closely linked to the locus for \*\*\*Xk\*\*\* [ \*\*\*protein\*\*\* ].

FILE 'MEDLINE'  
FILE 'JAPIO'  
FILE 'BIOSIS'  
FILE 'SCISEARCH'  
FILE 'WPIDS'  
FILE 'CAPLUS'  
FILE 'EMBASE'  
=> s xk protein#  
L1 74 XK PROTEIN#

=> I1 and ligand transport  
L2 0 L1 AND LIGAND TRANSPORT

=> I1 and (transport# or transporter# or transporting)  
L3 29 L1 AND (TRANSPORT# OR TRANSPORTER# OR TRANSPORTING)

=> dup rem I3  
PROCESSING COMPLETED FOR L3  
L4 17 DUP REM L3 (12 DUPLICATES REMOVED)

=> dup rem I1  
PROCESSING COMPLETED FOR L1  
L5 36 DUP REM L1 (38 DUPLICATES REMOVED)

=> d I5 ibib abs 1-36

L5 ANSWER 1 OF 36 MEDLINE  
ACCESSION NUMBER: 2003138455 MEDLINE  
DOCUMENT NUMBER: 22540026 PubMed ID: 12652714  
TITLE: Cellulitis, sepsis, acute renal failure and hemolytic anemia with McLeod blood group phenotype.  
AUTHOR: Furiya Shino; Kitazawa Kunihiko; Ideura Gen; Toshida Fumitaka; Shimizu Shinsuke; Shimajo Takashi; Sakai Toshiaki; Ishiguro Jun; Miyahara Takashige; Misawa Takuo; Noguchi Osamu  
CORPORATE SOURCE: Department of Internal Medicine, Nagano Matsushiro General Hospital, Nagano.  
SOURCE: NIPPON NAIKA GAKKAI ZASSHI. JOURNAL OF JAPANESE SOCIETY OF INTERNAL MEDICINE, (2003 Jan 10) 92 (1) 140-2. Journal code: 19130210R. ISSN: 0021-5384.  
PUB. COUNTRY: Japan  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: Japanese  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200305  
ENTRY DATE: Entered STN: 20030326  
Last Updated on STN: 20030521  
Entered Medline: 20030520

L5 ANSWER 2 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.  
ACCESSION NUMBER: 2003:235497 BIOSIS  
DOCUMENT NUMBER: PREV200300235497  
TITLE: An attempt at analysing the selected traits of body conformation, growth, performance and genetic structure of Lithuanian native Zemaitukai horse, the breed being preserved from extinction.  
AUTHOR(S): Macijauskiene, Vale (1); Juras, Rytis (1)  
CORPORATE SOURCE: (1) Lithuanian Institute of Animal Science, R. Zebenkos 12, Baisogala, LT-5125, Radviliskio Raj., Lithuania Lithuania  
SOURCE: Animal Science Papers and Reports, (2003) Vol. 21, No. 1, pp. 35-46. print. ISSN: 0860-4037.  
DOCUMENT TYPE: Article  
LANGUAGE: English  
AB Over the last 50 years the traits of valuable indigenous Lithuanian Zemaitukai horse have not been investigated and during the last decade the breed became on the verge of extinction. Recently certain measures were undertaken to preserve the breed, evaluate its present characteristics and compare them with those reported earlier. Body size and conformation traits of present-day Zemaitukai horse (ZH) were found corresponding or similar to those of the ancient type, showing that many valuable characteristics of the breed are retained. Mares' milk yield and composition were evaluated, as well as development of foals based on body dimensions. Genetic variation, genetic structure as well as relationship between the lines and families of ZH were studied using blood typing and electrophoretic analysis of serum proteins. Gene frequencies are presented at six blood group (A, D, C, Q, P, K) and five protein (AI, Es, Gc, XK, Tf) loci. The genetic diversity within blood groups and serum proteins in ZH kept in a closed population showed that out of eleven genetic systems examined, six were polymorphic. This is especially so for the A and D, as well as Es and Tf systems. The distribution of allele frequencies varied between the lines and families.

L5 ANSWER 3 OF 36 CAPLUS COPYRIGHT 2003 ACS  
ACCESSION NUMBER: 2002:755080 CAPLUS  
DOCUMENT NUMBER: 137:274161  
TITLE: Protein, gene and cDNA sequences of a novel human transport protein related to \*\*\*XK\*\*\*  
\*\*\*protein\*\*\* and their uses in drug screening  
INVENTOR(S): Merkulov, Gennady; Guegler, Karl; Brandon, Rhonda C.; Di Francesco, Valentina; Beasley, Ellen M.  
PATENT ASSIGNEE(S): USA  
SOURCE: U.S. Pat. Appl. Publ., 53 pp., Cont.-in-part of U.S. Ser. No. 740,034, abandoned. CODEN: USXXCO  
DOCUMENT TYPE: Patent  
LANGUAGE: English  
FAMILY ACC. NUM. COUNT: 2  
PATENT INFORMATION:

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
US 2002142376	A1	20021003	US 2001-768781	20010125
WO 2002072831	A2	20020919	WO 2002-US929	20020115
W:	AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, OM, PH, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TN, TR, TT, TZ, UA, UG, UZ, VN, YU, ZA, ZM, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM			
RW:	GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW, AT, BE, BG, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG			
PRIORITY APPLN. INFO.:			US 2000-740034 B2 20001220	
			US 2001-768781 A 20010125	
AB	The invention provides protein, cDNA and genomic sequences for a novel human transport protein XK. The transport protein gene is expressed in human germinal center B cell. Eight single nucleotide polymorphism has been found on transport protein XK gene mapped to chromosome 23. The invention also relates to screening modulator of transport protein XK and use them in therapy. The invention further relates to methods, vector and hosts for expression of transport protein XK.			

L5 ANSWER 4 OF 36 MEDLINE  
ACCESSION NUMBER: 2002480986 MEDLINE  
DOCUMENT NUMBER: 22228575 PubMed ID: 12243006  
TITLE: [Progress in molecular chorea diagnosis. McLeod syndrome and chorea acanthocytosis]. Fortschritte in der molekularen Chorea-Diagnostik. McLeod-Syndrom und Chorea-Akanthozytose.  
AUTHOR: Danek A  
CORPORATE SOURCE: Neurologische Klinik, Ludwig-Maximilians-Universitat, Postfach 701260, 81366 Munchen.. danek@nefro.med.uni-muenchen.de  
SOURCE: NERVENARZT, (2002 Jun) 73 (6) 564-9. Journal code: 0400773. ISSN: 0028-2804.  
PUB. COUNTRY: Germany: Germany, Federal Republic of  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: German  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200212  
ENTRY DATE: Entered STN: 20020924  
Last Updated on STN: 20021218  
Entered Medline: 20021217  
AB McLeod syndrome and chorea-acanthocytosis are classified with the so-called neuroacanthocytosis group of syndromes. Both lead to progressive basal ganglia degeneration and were not easily distinguished in the past. With the discovery of their molecular bases, mutations of the X-linked gene XK and autosomal recessive mutations of the gene coding for chorein, respectively, the two phenotypes can now be differentiated and extend the diagnostic spectrum in patients presenting with chorea. The present review compares the two conditions and proposes a practical approach to diagnosis and treatment. Better-defined disease concepts should eventually replace the umbrella term of "neuroacanthocytosis." Animal models are needed to understand the underlying mechanisms. A common pathway is likely for the pathogenesis of these conditions and is most probably shared with Huntington's disease.

L5 ANSWER 5 OF 36 CAPLUS COPYRIGHT 2003 ACS  
ACCESSION NUMBER: 2002:359541 CAPLUS  
DOCUMENT NUMBER: 137:214885  
TITLE: Point mutations causing the McLeod phenotype  
AUTHOR(S): Russo, David C. W.; Lee, Soohae; Reid, Marion E.;

Redman, Colvin M.  
CORPORATE SOURCE: The New York Blood Center, Lindsley F. Kimball Research Institute, New York, NY, USA  
SOURCE: Transfusion (Malden, MA, United States) (2002), 42(3), 287-293  
CODEN: TRANAT; ISSN: 0041-1132  
PUBLISHER: Blackwell Publishing, Inc.  
DOCUMENT TYPE: Journal  
LANGUAGE: English  
AB The McLeod phenotype is defined by absence of Kx, weakening of Kell system antigens, and acanthocytosis. Individuals with the McLeod phenotype usually develop late-onset neuromuscular abnormalities. Gene deletions, insertions, and point mutations that affect RNA splicing or that lead to premature stop codons were reported to cause the McLeod phenotype. The McLeod phenotype may also be caused by mutations at a different splice site and by a novel mutation encoding an amino acid substitution that prevents transport to the cell surface. The coding and flanking intron regions of XK from 4 male, unrelated individuals with the McLeod phenotype and nonchronic granulomatous disease were sequenced and compared with the wild type sequence. Genomic DNA was amplified by PCR, and the products were sequenced. In 1 case, the mutant cDNA was expressed in a heterologous cell, and cell surface expression was detd. 3 Individuals with the McLeod phenotype had mutations that disrupted conserved GT sequences present at RNA splice sites. 2 Of them had G>C mutations at the 5' splice site of intron 1, and 1 had a G>A mutation at the 5' splice site of intron 2. One person with the McLeod phenotype had a 746C>G mutation in exon 3 encoding an R222G substitution. In a transfected cell, the expressed protein from the latter mutant did not travel to the cell surface. The McLeod phenotype may be caused by several different mutations.  
REFERENCE COUNT: 25 THERE ARE 25 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L5 ANSWER 6 OF 36 EMBASE COPYRIGHT 2003 ELSEVIER SCI. B.V.  
ACCESSION NUMBER: 2002194888 EMBASE  
TITLE: [Differential diagnosis of hereditary chorea syndromes]. DIFFERENTIALDIAGNOSE HEREDITARER CHOREA-SYNDROME.  
AUTHOR: Jung H.H.  
CORPORATE SOURCE: Dr. H.H. Jung, Neurologische Klinik, Universitatsspital, Frauenklinikstrasse 26, CH-8091 Zurich, Germany. hans.jung@nos.usz.ch  
SOURCE: Schweizer Archiv fur Neurologie und Psychiatrie, (2002) 153/4 (185-188). Refs: 15 ISSN: 0258-7661 CODEN: SANPE8  
COUNTRY: Switzerland  
DOCUMENT TYPE: Journal; General Review  
FILE SEGMENT: 005 General Pathology and Pathological Anatomy  
008 Neurology and Neurosurgery  
022 Human Genetics  
032 Psychiatry  
LANGUAGE: German  
SUMMARY LANGUAGE: English  
AB The clinical triad of hereditary chorea syndromes includes (1) choreoform involuntary movement disorder, (2) psychiatric symptoms, and (3) cognitive impairment. The most frequent hereditary chorea syndrome is Huntington's disease (HD). There are several phenocopies of Huntington's disease, such as the Huntington's disease-like neurodegenerative disorders type 1 and type 2 (HDLD), benign hereditary chorea (BHC), dentato-rubro-pallido-Lusyan atrophy (DRPLA), choreoacanthocytosis (CHAC), and McLeod syndrome (MLS). Huntington's disease is caused by an instable CAG trinucleotide expansion in the Huntington disease gene, and onset age and severity of symptoms depend on the number of CAG repeats. The physiological function of the gene product Huntingtin and the disease mechanisms are not fully elucidated yet. However, experimental data strongly suggest that induction of apoptosis through a caspase (cysteine aspartate-specific proteases)-dependent mechanism might be an important factor for the development of the striatal neurodegeneration. The HDLDs are more or less exact phenocopies of Huntington's disease. Two chromosomal localisations are described, and one responsible gene, Junctophilin-3, is identified. The BHC manifests as a pure chorea syndrome, without major psychiatric or cognitive impairment. The disease is located on chromosome 14, but the responsible gene has not yet been

identified. Apart from the Huntington's disease-like phenotype, DRPLA may manifest as a spinocerebellar ataxia, a progressive myoclonus epilepsy, or mixed phenotypes. DRPLA is caused by instable CAG expansions in Atrophin-1, whose physiological functions are not yet known. CHAC and MLS belong to the so-called neuroacanthocytosis syndromes. CHAC is an autosomal-recessive disorder characterised by a progressive chorea syndrome, perioral dyskinesias and mutilations, and - less frequently - an akinetic-rigid extrapyramidal syndrome and seizures. The responsible gene is located on chromosome 9, encoding chorein, a protein implicated in intracellular cell sorting. MLS is an X-linked multi-system disorder with haematological, neuromuscular, and CNS involvement. Haematologically, MLS is characterised by absent expression of the Kx erythrocyte antigen, weak expression of Kell antigens, acanthocytosis, and a compensated haemolytic state. Asymptomatic males have elevated serum creatine kinase levels, and are prone to develop neurological symptoms. Neuromuscular manifestations include myopathy, sensory-motor axonal neuropathy, and cardiomyopathy. CNS manifestations comprise a choreaiform movement disorder, neuropsychiatric abnormalities, and - less frequently - generalised seizures. MLS is caused by mutations of the XK gene encoding the \*\*\*XK\*\*\* protein\*\*\*, a putative membrane transport protein containing the Kx erythrocyte antigen. The \*\*\*XK\*\*\* protein\*\*\* is linked to the Kell glycoprotein by a single disulfide bond, probably forming a functional complex. The Kell protein is a member of the metalloproteinase family, and the \*\*\*XK\*\*\* protein\*\*\* has functional similarities to the CED-8 protein in nematodes, in which it controls the timing of apoptosis. These data strongly suggest an important role of the XK-Kell complex in striatal physiology. The advances in the molecular biology of hereditary chorea syndromes offer the possibility for a direct genetic analysis of affected individuals, and presymptomatic testing for individuals at risk. Although the genetic bases of some hereditary chorea syndromes are established, causal therapies are lacking. However, the rapidly accumulating knowledge will hopefully lead to the development of efficient therapies that might attenuate or even prevent these otherwise relentlessly progressive neurodegenerative disorders.

L5 ANSWER 7 OF 36 MEDLINE DUPLICATE 1  
 ACCESSION NUMBER: 2001423940 MEDLINE  
 DOCUMENT NUMBER: 21347986 PubMed ID: 11375401  
 TITLE: Molecular defects underlying the Kell null phenotype.  
 AUTHOR: Lee S; Russo D C; Reiner A P; Lee J H; Sy M Y; Telen M J; Judd W J; Simon P; Rodrigues M J; Chabert T; Poole J; Jovanovic-Szreniuc S; Levene C; Yahalom V; Redman C M  
 CORPORATE SOURCE: Lindsley F. Kimball Research Institute of the New York Blood Center, New York, New York 10021, USA.  
 CONTRACT NUMBER: HL54459 (NHLBI)  
 SOURCE: JOURNAL OF BIOLOGICAL CHEMISTRY, (2001 Jul 20) 276 (29) 27281-9.  
 Journal code: 2985121R. ISSN: 0021-9258.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200108  
 ENTRY DATE: Entered STN: 20010827  
 Last Updated on STN: 20030105  
 Entered Medline: 20010823  
 AB Expression of the Kell blood group system is dependent on two proteins, Kell and XK, that are linked by a single disulfide bond. Kell, a type II membrane glycoprotein, is a zinc endopeptidase, while XK, which has 10 transmembrane domains, is a putative membrane transporter. A rare phenotype termed Kell null (Ko) is characterized by the absence of Kell protein and Kell antigens from the red cell membrane and diminished amounts of \*\*\*XK\*\*\* protein\*\*\*. We determined the molecular basis of eight unrelated persons with Ko phenotypes by sequencing the coding and the intron-exon splice regions of KEL and, in some cases, analysis of mRNA transcripts and expression of mutants on the cell surface of transfected cells. Six subjects were homozygous: four with premature stop codons, one with a 5' splice site mutation, G to A, in intron 3, and one with an amino acid substitution (S676N) in exon 18. Two Ko persons with premature stop codons had identical mutations in exon 4 (R128Stop), another had a different mutation in exon 4 (C83Stop), and the fourth had a stop codon in exon 9 (Q348Stop). Two Ko persons were heterozygous for two

mutations. One had a 5' splice site mutation (G to A) in intron 3 of one allele that caused aberrant splicing and exon skipping, and the other allele had an amino acid substitution in exon 10 (S363N). The other heterozygote had the same amino acid substitution in exon 10 (S363N) in one allele and a premature stop codon in exon 6 (R192Stop) in the other allele. The S363N and S676N mutants, expressed in 293T cells, were retained in a pre-Golgi compartment and were not transported to the cell surface, indicating that these mutations inhibit trafficking. We conclude that several different molecular defects cause the Kell null phenotype.

L5 ANSWER 8 OF 36 MEDLINE DUPLICATE 2  
 ACCESSION NUMBER: 2001514880 MEDLINE  
 DOCUMENT NUMBER: 21446863 PubMed ID: 11562915  
 TITLE: Kell and XK immunohistochemistry in McLeod myopathy.  
 AUTHOR: Jung H H; Russo D; Redman C; Brandner S  
 CORPORATE SOURCE: Department of Neurology, University Hospital Zurich, 8091 Zurich, Switzerland.. hans.jung@nos.usz.ch  
 CONTRACT NUMBER: HL54459 (NHLBI)  
 SOURCE: MUSCLE AND NERVE, (2001 Oct) 24 (10) 1346-51.  
 Journal code: 7803146. ISSN: 0148-639X.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200110  
 ENTRY DATE: Entered STN: 20010920  
 Last Updated on STN: 20021022  
 Entered Medline: 20011025  
 AB The McLeod syndrome is an X-linked neuroacanthocytosis manifesting with myopathy and progressive chorea. It is caused by mutations of the XK gene encoding the \*\*\*XK\*\*\* protein\*\*\*, a putative membrane transport protein of yet unknown function. In erythroid tissues, XK forms a functional complex with the Kell glycoprotein. Here, we present an immunohistochemical study in skeletal muscle of normal controls and a McLeod patient with a XK gene point mutation (C977T) using affinity-purified antibodies against XK and Kell proteins. Histological examination of the affected muscle revealed the typical pattern of McLeod myopathy including type 2 fiber atrophy. In control muscles, Kell immunohistochemistry stained sarcoplasmic membranes. XK immunohistochemistry resulted in a type 2 fiber-specific intracellular staining that was most probably confined to the sarcoplasmic reticulum. In contrast, there was only a weak background signal without a specific staining pattern for XK and Kell in the McLeod muscle. Our results demonstrate that the lack of physiological XK expression correlates to the type 2 fiber atrophy in McLeod myopathy, and suggest that the \*\*\*XK\*\*\* protein\*\*\* represents a crucial factor for the maintenance of normal muscle structure and function.  
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L5 ANSWER 9 OF 36 MEDLINE  
 ACCESSION NUMBER: 2001695801 MEDLINE  
 DOCUMENT NUMBER: 21612357 PubMed ID: 11746618  
 TITLE: The chorea of McLeod syndrome.  
 AUTHOR: Danek A; Tison F; Rubio J; Oechsner M; Kalkreuth W; Monaco A P  
 CORPORATE SOURCE: Neurologische Klinik, Ludwig-Maximilians-Universitat, Munchen, Germany..  
 danek@brain.nfo.med.uni-muenchen.de  
 SOURCE: MOVEMENT DISORDERS, (2001 Sep) 16 (5) 882-9.  
 Journal code: 8610688. ISSN: 0885-3185.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200202  
 ENTRY DATE: Entered STN: 20011218  
 Last Updated on STN: 20021022  
 Entered Medline: 20020213  
 AB Among the movement disorders associated with acanthocytosis, McLeod syndrome (McKusick 314850) is the one that is best characterized on the molecular level. Its defining feature is low reactivity of Kell erythrocyte antigens. This is due to absence of membrane protein KX that forms a complex with the Kell protein. KX is coded for by the XK gene on the X-chromosome. We present six males (aged 29 to 60 years), with proven XK mutations, to discuss the chorea associated with McLeod syndrome. The movement disorder commonly develops in the fifth decade and is progressive. It affects the limbs, the trunk and the face. In addition to facial grimacing, involuntary vocalization can be present. In early stages there may only be some restlessness or slight involuntary distal movements of ankles and fingers. Lip-biting and facial tics seem more common in autosomal recessive choreoacanthocytosis linked to chromosome 9. This, together with the absence of dysphagia in McLeod syndrome, may

help in differential diagnosis. Recent findings suggest a role for the endothelin system of the striatum in the pathogenesis of McLeod syndrome.  
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L5 ANSWER 10 OF 36 MEDLINE DUPLICATE 3  
 ACCESSION NUMBER: 2002035542 MEDLINE  
 DOCUMENT NUMBER: 21597008 PubMed ID: 11761473  
 TITLE: McLeod neuroacanthocytosis: genotype and phenotype.  
 AUTHOR: Danek A; Rubio J P; Rampoldi L; Ho M; Dobson-Stone C; Tison F; Symmans W A; Oechsner M; Kalkreuth W; Watt J M; Corbett A J; Hamdalla H H; Marshall A G; Sutton I; Dotti M T; Malandrini A; Walker R H; Daniels G; Monaco A P  
 CORPORATE SOURCE: Neurologische Klinik, Ludwig-Maximilians-Universitat, Munchen, Germany..  
 danek@brain.nfo.med.uni-muenchen.de  
 SOURCE: ANNALS OF NEUROLOGY, (2001 Dec) 50 (6) 755-64.  
 Journal code: 7707449. ISSN: 0364-5134.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200201  
 ENTRY DATE: Entered STN: 20020124  
 Last Updated on STN: 20021022  
 Entered Medline: 20020107  
 AB McLeod syndrome is caused by mutations of XK, an X-chromosomal gene of unknown function. Originally defined as a peculiar Kell blood group variant, the disease affects multiple organs, including the nervous system, but is certainly underdiagnosed. We analyzed the mutations and clinical findings of 22 affected men, aged 27 to 72 years. Fifteen different XK mutations were found, nine of which were novel, including the one of the eponymous case McLeod. Their common result is predicted absence or truncation of the \*\*\*XK\*\*\* protein\*\*\*. All patients showed elevated levels of muscle creatine phosphokinase, but clinical myopathy was less common. A peripheral neuropathy with areflexia was found in all but 2 patients. The central nervous system was affected in 15 patients, as obvious from the occurrence of seizures, cognitive impairment, psychopathology, and choreatic movements. Neuroimaging emphasized the particular involvement of the basal ganglia, which was also detected in 1 asymptomatic young patient. Most features develop with age, mainly after the fourth decade. The resemblance of McLeod syndrome with Huntington's disease and with autosomal recessive chorea-acanthocytosis suggests that the corresponding proteins--XK, huntingtin, and chorein--might belong to a common pathway, the dysfunction of which causes degeneration of the basal ganglia.

L5 ANSWER 11 OF 36 MEDLINE DUPLICATE 4  
 ACCESSION NUMBER: 2001161085 MEDLINE  
 DOCUMENT NUMBER: 21157963 PubMed ID: 11261514  
 TITLE: McLeod syndrome: a novel mutation, predominant psychiatric manifestations, and distinct striatal imaging findings.  
 AUTHOR: Jung H H; Hergersberg M; Kneifet S; Alkadhi H; Schiess R; Weigell-Weber M; Daniels G; Kollias S; Hess K  
 CORPORATE SOURCE: Department of Neurology, University Hospital Zurich, Switzerland.. hans.jung@nos.usz.ch  
 SOURCE: ANNALS OF NEUROLOGY, (2001 Mar) 49 (3) 384-92.  
 Journal code: 7707449. ISSN: 0364-5134.  
 PUB. COUNTRY: United States  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 200104  
 ENTRY DATE: Entered STN: 20010425  
 Last Updated on STN: 20010425  
 Entered Medline: 20010419  
 AB The McLeod syndrome is an X-linked disorder caused by mutations of the XK gene encoding the \*\*\*XK\*\*\* protein\*\*\*. The syndrome is characterized by absent Kx erythrocyte antigen, weak expression of Kell blood group system antigens, and acanthocytosis. In some allelic variants, elevated creatine kinase, myopathy, neurogenic muscle atrophy, and progressive chorea are found. We describe a family with a novel point mutation in the XK gene consisting of a C to T base transition at nucleotide position 977, introducing a stop codon. Among seven affected males, five manifested with psychiatric disorders such as depression, bipolar disorder, or personality disorder, but only two presented with chorea. Positron emission tomography and magnetic resonance volumetry revealed reduced striatal 2-fluoro-2-deoxy-glucose (FDG) uptake and

diminished volumes of the caudate nucleus and putamen that correlated with disease duration. In contrast, none of 12 female mutation carriers showed psychiatric or movement disorders. However, a semidominant effect of the mutation was suggested by erythrocyte and blood group mosaicism and reduced striatal FDG uptake without structural abnormalities. Therefore, patients with psychiatric signs or symptoms segregating in an X-linked trait should be examined for acanthocytosis and Kell/Kx blood group serology.

L5 ANSWER 12 OF 36 MEDLINE DUPLICATE 5  
ACCESSION NUMBER: 2001674195 MEDLINE  
DOCUMENT NUMBER: 21560274 PubMed ID: 11703337  
TITLE: A spontaneous novel XK gene mutation in a patient with McLeod syndrome.  
AUTHOR: Supple S G; Iland H J; Barnett M H; Pollard J D  
CORPORATE SOURCE: The Kanematsu Laboratories, Royal Prince Alfred Hospital, Camperdown, NSW, Australia.  
SOURCE: BRITISH JOURNAL OF HAEMATOLOGY, (2001 Nov) 115 (2) 369-72.  
Journal code: 0372544. ISSN: 0007-1048.  
PUB. COUNTRY: England; United Kingdom  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200112  
ENTRY DATE: Entered STN: 20011127  
Last Updated on STN: 20021022  
Entered Medline: 20011207  
AB A 29-year-old man with a history of elevated creatine kinase and necrotizing myopathy was reviewed. Prominent red cell acanthocytosis in association with reduced Kell antigen expression was present, findings consistent with the McLeod syndrome. Investigation of the patient's XK gene revealed a novel TGG- to-TAG transition at position 1023 in exon 3. This point mutation creates an in-frame stop codon (W314X), and predicts a truncated \*\*\*XK\*\*\* \*\*\*protein\*\*\* of 313 amino acids, compared with 444 amino acids in the normal \*\*\*XK\*\*\* \*\*\*protein\*\*\*. The mutation was not identified in the patient's mother or sister indicating that this mutation was spontaneous.

L5 ANSWER 13 OF 36 CAPLUS COPYRIGHT 2003 ACS  
ACCESSION NUMBER: 2001:934288 CAPLUS  
DOCUMENT NUMBER: 136:115930  
TITLE: Use of blood protein polymorphism for determining genetic distance between half-bred stallions  
AUTHOR(S): Pikula, Ryszard; Tomaszewska-Guszkiewicz, Krystyna;  
Smugala, Mirosław; Gronet, Dominik  
CORPORATE SOURCE: Dep. of Horse Breeding, Agricultural Univ. of Szczecin, Szczecin, 71-466, Pol.  
SOURCE: Folia Universitatis Agriculturae Stetinensis (2001), 219, 67-71  
CODEN: FUASFI; ISSN: 1506-1965  
PUBLISHER: Wydawnictwo Akademii Rolniczej w Szczecinie  
DOCUMENT TYPE: Journal  
LANGUAGE: English  
AB Genetic blood protein polymorphism of stallions was used to describe genetically 3 breeds of half-bred horses. The investigations covered Malopolski, Wielkopolski, and noble half-bred stallions from which blood samples were collected; in the samples, polymorphism of selected proteins: albumin (Al), transferrin (Tf), 8.5 pH esterase (EspH 8.5), vitamin D-binding protein (Gc), and \*\*\*Xk\*\*\* \*\*\*protein\*\*\* (Xk), was detd. On the grounds of the performed studies, significant differences were found in phenotypic and allelic frequencies of blood protein systems analyzed according to the stallion breed. The av. heterozygosity and homozygosity coeffs. were established for stallion breeds as well as genetic similarity and genetic distance between breeds of the stallions. This distance was: 0.01046 between Malopolski and Wielkopolski stallions, 0.01783 between Malopolski and noble half-bred stallions, and 0.01000 between Wielkopolski and noble half-bred stallions.  
REFERENCE COUNT: 7 THERE ARE 7 CITED REFERENCES  
AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L5 ANSWER 14 OF 36 MEDLINE DUPLICATE 6  
ACCESSION NUMBER: 2001063593 MEDLINE  
DOCUMENT NUMBER: 20533666 PubMed ID: 11099667  
TITLE: First example of anti-Kx in a person with the McLeod phenotype and without chronic granulomatous disease.  
AUTHOR: Russo D C; Oyen R; Powell V I; Perry S; Hitchcock J; Redman C M; Reid M E  
CORPORATE SOURCE: New York Blood Center, New York, New York, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: TRANSFUSION, (2000 Nov) 40 (11) 1371-5.  
Journal code: 0417360. ISSN: 0041-1132.

PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200012  
ENTRY DATE: Entered STN: 20010322  
Last Updated on STN: 20010322  
Entered Medline: 20001222  
AB BACKGROUND: Kx is lacking in the RBCs of patients with the McLeod syndrome. This condition is sometimes associated with chronic granulomatous disease (CGD). If given allogeneic RBCs, CGD patients with the McLeod phenotype may produce anti-Kx and anti-Km, and only phenotypically matched McLeod blood would be compatible. McLeod phenotype persons without CGD have made anti-Km but not anti-Kx (2 examples), and thus both McLeod and K(O) blood would be compatible. CASE REPORT: RBCs from a transfused patient with the McLeod phenotype but not with CGD (non-CGD McLeod) were typed for the Kell blood group antigens, and the plasma was analyzed for the presence of antibody by agglutination. The molecular basis was determined by analyzing for \*\*\*XK\*\*\* \*\*\*protein\*\*\* on RBC membranes by Western immunoblotting, by sequencing the XK gene, and by RFLP. RESULTS: The RBCs did not react with anti-Kx + anti-Km and showed weakening of Kell system antigens. The patient's plasma reacted moderately (2+) with RBCs of common Kell type and strongly (4+) with K(O) RBCs and RBCs of common Kell type treated with dithiothreitol, and did not react with McLeod RBCs. \*\*\*XK\*\*\* \*\*\*protein\*\*\* was absent from the RBC membranes. The XK gene had a point mutation in the donor splice site of intron 1 (G>C).  
CONCLUSION: This is the first report describing the molecular alteration in a non-CGD McLeod patient who has made anti-Kx. The immune response of people with the McLeod phenotype can vary, and K(O) blood may not always be compatible.

L5 ANSWER 15 OF 36 MEDLINE DUPLICATE 7  
ACCESSION NUMBER: 2000384103 MEDLINE  
DOCUMENT NUMBER: 20352021 PubMed ID: 10891471  
TITLE: Expression of Kell blood group protein in nonerythroid tissues.  
AUTHOR: Russo D; Wu X; Redman C M; Lee S  
CORPORATE SOURCE: Lindsley F. Kimball Research Institute, The New York Blood Center, New York, New York 10021, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: BLOOD, (2000 Jul 1) 96 (1) 340-6.  
Journal code: 7603509. ISSN: 0006-4971.  
PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Abridged Index Medicus Journals; Priority Journals  
ENTRY MONTH: 200008  
ENTRY DATE: Entered STN: 20000818  
Last Updated on STN: 20021022  
Entered Medline: 20000810  
AB The Kell blood group protein is a zinc endopeptidase that yields endothelin-3, a potent bioactive peptide, by cleavage of big endothelin-3, a larger intermediate precursor. On red cells, Kell protein is linked by a single disulfide bond to XK, a protein that traverses the membrane 10 times and whose absence, as occurs in the McLeod phenotype, is associated with a set of clinical symptoms that include nerve and muscle disorders and red cell acanthocytosis. Previous studies indicated that Kell is primarily expressed in erythroid tissues, whereas XK has a wider tissue distribution. The tissue distribution of Kell protein has been further investigated by Northern blot analysis, PCR-screening of tissue complementary DNAs (cDNAs), and Western immunoblots. Screening of an RNA dot-blot panel confirmed that Kell is primarily expressed in erythroid tissues but is also expressed in a near equal amount in testis, with weaker expression in a large number of other tissues. PCR-screening of cDNAs from different tissues and DNA sequencing of the products gave similar results. In 2 of the nonerythroid tissues tested, testis and skeletal muscle, Kell protein was detected by Western immunoblotting. In skeletal muscle, isolation of XK with a specific antibody coisolated Kell protein. These studies demonstrate that Kell is expressed in both erythroid and nonerythroid tissues and is associated with XK.

L5 ANSWER 16 OF 36 MEDLINE  
ACCESSION NUMBER: 2000384510 MEDLINE  
DOCUMENT NUMBER: 20307454 PubMed ID: 10849386  
TITLE: A murine monoclonal antibody against Kx protein which reacts also with beta-spectrin.  
AUTHOR: Carbonnet F; Blanchard D; Hattab C; Cochet S; Petit-Leroux Y; Loirat M J; Cartron J P; Bertrand O  
CORPORATE SOURCE: INSERM U76, Institut National de la Transfusion Sanguine, Alexandre Cabanel, Paris, France.

SOURCE: TRANSFUSION MEDICINE, (2000 Jun) 10 (2) 145-54.  
Journal code: 9301182. ISSN: 0958-7578.  
PUB. COUNTRY: ENGLAND; United Kingdom  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200008  
ENTRY DATE: Entered STN: 20000818  
Last Updated on STN: 20021022  
Entered Medline: 20000808  
AB Kx is a polytopic membrane protein of human erythrocytes carrying the Kx blood group antigen, which is deficient in rare patients with McLeod syndrome. Kx is disulphide bond linked to the Kell glycoprotein, which is a bitopic type II membrane protein carrying the Kell blood group antigen. Mice immunized with a synthetic peptide predicted to be located on the second external loop of Kx produced a monoclonal antibody called 3E12 which does not recognize red cells with common Kell phenotype by agglutination and flow cytometry. 3E12 recognizes the Kx protein and the spectrin beta-chain on western blots, the affinity for these two proteins being lowered with increasing ionic strength. Linear epitopes recognized by 3E12 are E116IEIKE121 and L484AQELEKE491 on the Kx protein and spectrin beta-chain, respectively. To quantify the relative amount of Kx in Empigen BB extracts of red cell membranes, an ELISA for Kx was set up which showed conclusively that (i) there is less Kx in membranes of K0 individuals (lacking the Kell glycoprotein) than in membranes of common individuals, and (ii) that all common individuals, typed as K+k-, K-k+ and K+k+, have the same amount of Kx on their red cell membranes. When an erythrocyte membrane detergent extract from one K0 individual was chromatographed on an immobilized 3E12 column, a minute amount of authentic Kell glycoprotein was recovered in acid eluted fractions, indicating that at least the K0 individual under study may still produce some Kell protein.

L5 ANSWER 17 OF 36 MEDLINE DUPLICATE 8  
ACCESSION NUMBER: 2000250542 MEDLINE  
DOCUMENT NUMBER: 20250542 PubMed ID: 10791880  
TITLE: The Kell blood group system: Kell and XK membrane proteins.  
AUTHOR: Lee S; Russo D; Redman C M  
CORPORATE SOURCE: Lindsley F. Kimball Research Institute, The New York Blood Center, New York 10021, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: SEMINARS IN HEMATOLOGY, (2000 Apr) 37 (2) 113-21. Ref: 62  
Journal code: 0404514. ISSN: 0037-1963.  
PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
General Review; (REVIEW)  
(REVIEW, TUTORIAL)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 200006  
ENTRY DATE: Entered STN: 20000629  
Last Updated on STN: 20021022  
Entered Medline: 20000621  
AB Two membrane proteins express the antigens that comprise the Kell blood group system. A single antigen, Kx, is carried on XK, a 440-amino acid protein that spans the membrane 10 times, and more than 20 antigens reside on Kell, a 93-kd, type II glycoprotein. XK and Kell are linked, close to the membrane surface, by a single disulfide bond between Kell cysteine 72 and XK cysteine 347. Although primarily expressed in erythroid tissues, Kell and XK are also present in many other tissues. The polymorphic forms of Kell are due to single base mutations that encode different amino acids. Some Kell antigens are highly immunogenic and may cause strong reactions if mismatched blood is transfused and severe fetal anemia in sensitized mothers. Antibodies to KEL1 may suppress erythropoiesis at the progenitor level, leading to fetal anemia. The cellular functions of Kell/XK are complex. Absence of XK, the McLeod phenotype, is associated with acanthocytic red blood cells (RBCs), and with late-onset forms of muscular dystrophy and nerve abnormalities. Kell, by homology, is a member of the nephrilysin (M13) family of membrane zinc endopeptidases and it preferentially activates endothelin-3 by specific cleavage of the Trp21-Ile22 bond of big endothelin-3.

L5 ANSWER 18 OF 36 MEDLINE DUPLICATE 9  
ACCESSION NUMBER: 2000244352 MEDLINE  
DOCUMENT NUMBER: 20244352 PubMed ID: 10782495  
TITLE: Functional and structural aspects of the Kell blood group system.  
AUTHOR: Lee S; Russo D; Redman C

CORPORATE SOURCE: Lindsley F Kimball Research Institute of the New York Blood

Center, NY 10021, USA.

CONTRACT NUMBER: HL54459 (NHLBI)

SOURCE: TRANSFUSION MEDICINE REVIEWS, (2000 Apr) 14 (2) 93-103.

Ref: 49

Journal code: 8709027. ISSN: 0887-7963.

PUB. COUNTRY: United States

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

General Review; (REVIEW)

(REVIEW, TUTORIAL)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 200006

ENTRY DATE: Entered STN: 20000629

Last Updated on STN: 20021022

Entered Medline: 20000616

AB Two covalently linked proteins, Kell and XK, constitute the Kell blood

group system. Kell, a 93-kDa type II glycoprotein, is highly polymorphic and carries all but 1 of the known Kell antigens, and XK, which traverses

the membrane 10 times, carries a single antigen, the ubiquitous Kx. The Kell/XK complex is not limited to erythroid tissues and may have multiple

physiological roles. Absence of one of the component proteins, XK, is associated with abnormal red cell morphology and late-onset forms of nerve

and muscle abnormalities, whereas the other protein component, Kell, is an

enzyme whose principal known function is the production of a potent bioactive peptide, ET-3.

L5 ANSWER 19 OF 36 MEDLINE DUPLICATE 10

ACCESSION NUMBER: 2001085975 MEDLINE

DOCUMENT NUMBER: 21015021 PubMed ID: 11132157

TITLE: The mouse Kell blood group gene (Kel): cDNA sequence, genomic organization, expression, and enzymatic function.

AUTHOR: Lee S; Russo D C; Pu J; Ho M; Redman C M

CORPORATE SOURCE: The Lindsley F. Kimball Research Institute of the New York

Blood Center, NY 10021, USA.

CONTRACT NUMBER: HL54459 (NHLBI)

SOURCE: IMMUNOGENETICS, (2000 Nov) 52 (1-2) 53-62.

Journal code: 0420404. ISSN: 0093-7711.

PUB. COUNTRY: United States

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Priority Journals

OTHER SOURCE: GENBANK-AF252870

ENTRY MONTH: 200101

ENTRY DATE: Entered STN: 20010322

Last Updated on STN: 20021022

Entered Medline: 20010118

AB The human Kell blood group system is important in transfusion medicine,

since Kell is a polymorphic protein and some of its antigens can cause severe reactions if mismatched blood is transfused, while maternal alloimmunization may lead to fetal and neonatal anemia. In humans, Kell

is an Mr 93,000 type II membrane glycoprotein with endothelin-3-converting

enzyme activity that is linked by a single disulfide bond to another protein, XK, that spans the membrane ten times. An absence of XK leads to

clinical symptoms termed the McLeod syndrome. We determined the cDNA

sequence of the mouse Kell homologue, the organization of the gene, expression of the protein and its enzymatic function on red cells. Comparison of human and mouse Kell cDNA showed 80% nucleotide and 74%

amino acid sequence identity. Notable differences are that the mouse Kell

protein has eight probable N-linked carbohydrate side chains, compared to

five for human Kell, and that the mouse homologue has one more extracellular cysteine than human Kell protein. The mouse Kell gene (Kel), like its human counterpart, is similarly organized into 19 exons. Kel was located to proximal Chromosome 6. Northern blot analysis showed

high expression in spleen and weaker levels in testis and heart. Western blot analysis of red cell membrane proteins demonstrated that mouse

Kell glycoprotein has an apparent Mr of 110,000 and, on removal of

N-linked sugars, 80,000. As in human red cells, Kell is disulfide-linked to XK and

mouse red cells have endothelin-3-converting enzyme activity.

L5 ANSWER 20 OF 36 MEDLINE

ACCESSION NUMBER: 2000411485 MEDLINE

DOCUMENT NUMBER: 20353811 PubMed ID: 10895256

TITLE: Kell, Kx and the McLeod syndrome.

AUTHOR: Redman C M; Russo D; Lee S

CORPORATE SOURCE: Laboratory of Membrane Biochemistry, Lindsley F. Kimball

Research Institute, New York Blood Center, NY 10021,

USA..

credman@nybc.org

SOURCE: Baillieres Best Pract Res Clin Haematol, (1999 Dec) 12 (4)

621-35. Ref: 95

Journal code: 100900679. ISSN: 1521-6926.

PUB. COUNTRY: ENGLAND: United Kingdom

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

General Review; (REVIEW)

(REVIEW, TUTORIAL)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 200008

ENTRY DATE: Entered STN: 20000907

Last Updated on STN: 20021022

Entered Medline: 20000829

AB The antigens of the Kell blood group system are carried on a 93 kDa type

II glycoprotein encoded by a single gene on chromosome 7 at 7q33.

XK is a

50.9 kDa protein that traverses the membrane ten times and derives from a

single gene on the X chromosome at Xp21. A single disulphide bond, Kell

Cys 72-XK Cys 347, links Kell to XK. The Kell component of the Kell/XK

complex is important in transfusion medicine since it is a highly polymorphic protein, carrying over 23 different antigens, that can cause severe reactions if mismatched blood is transfused and in pregnant

mothers

antibodies to Kell may elicit serious fetal and neonatal anaemia. The different Kell phenotypes are all caused by base mutations leading to single amino acid substitutions. By contrast the XK component carries

a

single blood group antigen, termed Kx. The physiological functions of Kell and XK have not been fully elucidated but Kell is a zinc

endopeptidase with endothelin-3-converting enzyme activity and XK has the

structural characteristics of a membrane transporter. Lack of Kx, the McLeod phenotype, is associated with red cell acanthocytosis, elevated levels of serum creatine phosphokinase and late onset forms of muscular and neurological defects.

L5 ANSWER 21 OF 36 MEDLINE

ACCESSION NUMBER: 200009522 MEDLINE

DOCUMENT NUMBER: 20009522 PubMed ID: 10541802

TITLE: Structure and expression of the mouse homologue of the XK

gene.

AUTHOR: Collee E; Colin Y; Carbonnet F; Hattab C; Bertrand O; Cartton J P; Kim C L

CORPORATE SOURCE: INSERM U76, Institut National de la Transfusion Sanguine, 6

rue Alexandre Cabanel, 75015 Paris, France.

SOURCE: IMMUNOGENETICS, (1999 Oct) 50 (1-2) 16-21.

Journal code: 0420404. ISSN: 0093-7711.

PUB. COUNTRY: United States

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Priority Journals

OTHER SOURCE: GENBANK-AF155511

ENTRY MONTH: 199911

ENTRY DATE: Entered STN: 20000111

Last Updated on STN: 20021022

Entered Medline: 19991119

AB The human Kx blood group antigen is carried by a 37,000 M(r) apparent

molecular mass membrane polypeptide which is deficient in rare individuals

with the McLeod syndrome. The X-linked human XK gene is transcribed in

many tissues including adult skeletal muscle and brain, sieges of disorders observed in McLeod syndrome. We report here the cloning of the

orthologous mouse XK mRNA. Comparison of XK from human and mouse revealed

80% sequence similarity at the amino acid level. The mouse XK gene is organized in two exons and is expressed in many tissues, but its

expression pattern is slightly different from that of the human gene. The presence in mouse erythrocyte membrane of a 43,000 M(r) Kx-related

protein was demonstrated by immunoblotting with a rabbit antiserum directed against the human protein. With non-reduced samples, a 140,000 M(r)

species was detected instead of the 43,000 M(r) protein, suggesting that,

as demonstrated in the Kx polypeptide might be complexed with another

protein in mouse red cells, presumably the homologue of the human Kell

protein of 93,000 M(r).

L5 ANSWER 22 OF 36 MEDLINE DUPLICATE 11

ACCESSION NUMBER: 2000025439 MEDLINE

DOCUMENT NUMBER: 20025439 PubMed ID: 10556484

TITLE: Intracellular assembly of Kell and XK blood group proteins.

AUTHOR: Russo D; Lee S; Redman C

CORPORATE SOURCE: Lindsley F. Kimball Research Institute, The New York Blood

Center, 310 East 67 Street, New York, NY, USA.

CONTRACT NUMBER: HL54459 (NHLBI)

SOURCE: BIOCHIMICA ET BIOPHYSICA ACTA, (1999 Nov 9) 1461 (1) 10-18.

Journal code: 0217513. ISSN: 0006-3002.

PUB. COUNTRY: Netherlands

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 199912

ENTRY DATE: Entered STN: 20000113

Last Updated on STN: 20021022

Entered Medline: 19991227

AB Kell, a 93 kDa type II membrane glycoprotein, and XK, a 444 amino acid

multi-pass membrane protein, are blood group proteins that exist as a disulfide-bonded complex on human red cells. The mechanism of

Kell/XK

assembly was studied in transfected COS cells co-expressing Kell and \*\*\*XK\*\*\* \*\*\*proteins\*\*\*. Time course studies combined with

endonuclease-H treatment and cell fractionation showed that Kell and XK

are assembled in the endoplasmic reticulum. At later times the Kell component of the complex was not cleaved by endonuclease-H,

indicating

N-linked oligosaccharide processing and transport of the complex to a Golgi and/or a post-Golgi cell fraction. Surface-labeling of transfected

COS cells, expressing both Kell and XK, demonstrated that the Kell/XK complex travels to the plasma membrane. XK expressed in the absence

of

Kell was also transported to the cell surface indicating that linkage of

Kell and XK is not obligatory for cell surface expression.

L5 ANSWER 23 OF 36 SCISEARCH COPYRIGHT 2003 THOMSON ISI

ACCESSION NUMBER: 1999-939262 SCISEARCH

THE GENUINE ARTICLE: 260MH

TITLE: Intracellular assembly of Kell and XK blood group

proteins

AUTHOR: Russo D; Lee S; Redman C (Reprint)

CORPORATE SOURCE: NEW YORK BLOOD CTR, LINDSLEY F

KIMBALL RES INST, 310 E 67

ST, NEW YORK, NY 10021 (Reprint); NEW YORK

BLOOD CTR,

LINDSLEY F KIMBALL RES INST, NEW YORK, NY

10021

COUNTRY OF AUTHOR: USA

SOURCE: BIOCHIMICA ET BIOPHYSICA

ACTA-BIOMEMBRANES, (9 NOV 1999)

Vol. 1461, No. 1, pp. 10-18.

Publisher: ELSEVIER SCIENCE BV, PO BOX 211, 1000

AE

AMSTERDAM, NETHERLANDS.

ISSN: 0005-2736.

DOCUMENT TYPE: Article; Journal

FILE SEGMENT: LIFE

LANGUAGE: English

REFERENCE COUNT: 36

\*ABSTRACT IS AVAILABLE IN THE ALL AND IALL

FORMATS\*

AB Kell, a 93 kDa type II membrane glycoprotein, and XK, a 444

amino acid

multi-pass membrane protein, are blood group proteins that exist as a disulfide-bonded complex on human red cells. The mechanism of

Kell/XK

assembly was studied in transfected COS cells co-expressing Kell and \*\*\*XK\*\*\* \*\*\*proteins\*\*\*. Time course studies combined with

endonuclease-H treatment and cell fractionation showed that Kell and XK

are assembled in the endoplasmic reticulum. At later times the Kell component of the complex was not cleaved by endonuclease-H,

indicating

N-linked oligosaccharide processing and transport of the complex to a Golgi and/or a post-Golgi cell fraction. Surface-labeling of transfected

COS cells, expressing both Kell and XK, demonstrated that the Kell/XK complex travels to the plasma membrane. XK expressed in the absence

of

Kell was also transported to the cell surface indicating that linkage of

Kell and XK is not obligatory for cell surface expression. (C) 1999

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L5 ANSWER 24 OF 36 MEDLINE

ACCESSION NUMBER: 1999353182 MEDLINE

DOCUMENT NUMBER: 99353182 PubMed ID: 10426139

TITLE: A novel frameshift mutation in the McLeod syndrome

gene in

a Japanese family.

AUTHOR: Hanaoka N; Yoshida K; Nakamura A; Furihata K; Seo T; Tani

Y; Takahashi J; Ikeda S; Hanyu N

CORPORATE SOURCE: Department of Medicine (Neurology), Shinshu University

School of Medicine, Matsumoto, Japan.

SOURCE: JOURNAL OF THE NEUROLOGICAL SCIENCES,

(1999 May 1) 165 (1)

6-9.

Journal code: 0375403. ISSN: 0022-510X.

PUB. COUNTRY: Netherlands

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English

FILE SEGMENT: Priority Journals

ENTRY MONTH: 199909

ENTRY DATE: Entered STN: 19991005

Last Updated on STN: 20021022

Entered Medline: 19990920

AB We report a novel mutation in the XK gene (XK) in a Japanese patient with

McLeod syndrome. A 50-year-old man showed progressive muscular atrophy.

choreic movement, elevated level of serum creatinine kinase, and acanthocytosis. The expression level of all the Kell antigens in erythrocyte was decreased and molecular analysis revealed a single-base (T) deletion at the nucleotide position 1095 in XK. This deletion caused a frameshift in translation, leading to a premature stop codon at the amino acid position 408. We conclude this single-base deletion causes defective Kx protein, which is responsible for the McLeod phenotype in this patient.

L5 ANSWER 25 OF 36 MEDLINE DUPLICATE 12  
ACCESSION NUMBER: 1998256328 MEDLINE  
DOCUMENT NUMBER: 98256328 PubMed ID: 9593744  
TITLE: Association of XK and Kell blood group proteins.  
AUTHOR: Russo D; Redman C; Lee S  
CORPORATE SOURCE: Lindsley F. Kimball Research Institute, New York Blood Center, New York, New York 10021, USA.  
CONTRACT NUMBER: HL54459 (NHLBI)  
SOURCE: JOURNAL OF BIOLOGICAL CHEMISTRY, (1998 May 29) 273 (22) 13950-6.  
Journal code: 2985121R. ISSN: 0021-9258.

PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 199807  
ENTRY DATE: Entered STN: 19980713  
Last Updated on STN: 20021022  
Entered Medline: 19980701

AB A disulfide bond links Kell and XK red cell membrane proteins. Kell, a type II membrane glycoprotein, carries over 20 blood group antigens, and XK, which spans the membrane 10 times, is lacking in rare individuals with the McLeod syndrome. Kell is classified in the neprilysin family of zinc endopeptidases, and XK has structural features that suggest it is a transport protein. Kell has 15 extracellular cysteines, and XK has one

in its fifth extracellular loop. Five of the extracellular cysteine residues in Kell are not conserved in the other members of the neprilysin family, and based on the hypothesis that one of the nonconserved cysteines is linked to XK, cysteines 72 and 319 were mutated to serine. The single extracellular cysteine 347 of XK was also mutated. Co-expression of combinations of wild-type and mutant proteins in transfected COS-1 cells showed that Kell C72S did not form a Kell-XK complex with wild-type XK, while wild-type Kell and Kell C319S did. XK C347S was also unable to form a complex with wild-type Kell, indicating that Kell cysteine 72 is linked to XK cysteine 347. Kell C72S was transported to the cell surface, indicating that linkage to XK is not required. In addition, chemical cross-linking of red cell membranes with dithiobispropionimide indicated that glyceraldehyde-3-phosphate dehydrogenase is a near neighbor of Kell.

L5 ANSWER 26 OF 36 MEDLINE  
ACCESSION NUMBER: 1999003496 MEDLINE  
DOCUMENT NUMBER: 99003496 PubMed ID: 9784384  
TITLE: Kx, a quantitatively minor protein from human erythrocytes, is palmitoylated in vivo.  
AUTHOR: Carbonnet F; Hattab C; Callebaut I; Cochet S; Blancher A;  
Cartron J P; Bertrand O  
CORPORATE SOURCE: Institut National de la Transfusion Sanguine, 6 rue Alexandre Cabanel, Paris, 75015, France.  
SOURCE: BIOCHEMICAL AND BIOPHYSICAL RESEARCH COMMUNICATIONS, (1998 Sep 29) 250 (3) 569-74.  
Journal code: 0372516. ISSN: 0006-291X.

PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
ENTRY MONTH: 199811  
ENTRY DATE: Entered STN: 19990106  
Last Updated on STN: 20021022  
Entered Medline: 19981123

AB Kx is a quantitatively minor blood group protein of human erythrocytes which is thought to be a membrane transporter. In the red cell membrane, Kx forms a complex stabilized by a disulfide bond with the Kell blood group membrane protein which might function as a metalloprotease. The palmitoylation status of these proteins was studied by incubating red cells with [3H] palmitic acid. Purification of the Kell-Kx complex, by immunochromatography on an immobilized human monoclonal antibody of Kell blood group specificity demonstrated that the Kx but not the Kell protein is palmitoylated. Six cysteines in Kx are predicted to be intracytoplasmic and might be targets for palmitoylation. Three of these cysteines are present in a portion of sequence which is predicted to form an amphipathic alpha helix. Palmitoylation of one or several of these cysteines might contribute to anchor the cytoplasmic portion of the Kx

protein to the inner surface of red cell membrane.  
Copyright 1998 Academic Press.

L5 ANSWER 27 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.  
ACCESSION NUMBER: 2000:267961 BIOSIS  
DOCUMENT NUMBER: PREV200000267961  
TITLE: Genetic analysis in the Basque Pony-Pottoka breed: Preliminary results.  
AUTHOR(S): Pascual Moro, I. (1); Tejedor, T.; Monteagudo Ibanez, L. V.;  
Intxausti del Casal, J. I. (1); Arruga Lavina, M. V.  
CORPORATE SOURCE: (1) Servicio de Ganaderia, Diputacion Foral de Bizkaia,  
Lehendakari Agirre Etorbidea, 9, 2, 48014, Bilbao Spain  
SOURCE: Archivos de Zootecnia, (1998) Vol. 47, No. 178-179, pp.  
181-188.  
Meeting Info.: Spanish Society for the animal Genetic Resources. Cordoba, Spain December 14-17, 1997 Nacional de la Sociedad Espanola para los Recursos Geneticos Animales . ISSN: 0004-0592.  
DOCUMENT TYPE: Conference  
LANGUAGE: Spanish  
SUMMARY LANGUAGE: English

L5 ANSWER 28 OF 36 SCISEARCH COPYRIGHT 2003 THOMSON ISI  
ACCESSION NUMBER: 97:609377 SCISEARCH  
THE GENUINE ARTICLE: XQ325  
TITLE: Analysis of the McLeod syndrome gene in three patients with neuroacanthocytosis  
AUTHOR: Shizuka M; Watanabe M; Aoki M; Ikeda Y; Mizushima K;  
Okamoto K; Itoyama Y; Abe K; Shoji M (Reprint)  
CORPORATE SOURCE: GUNMA UNIV, SCH MED, DEPT NEUROL, 3-39-15 SHOWA MACHI, MAEBASHI, GUMMA 371, JAPAN (Reprint); GUNMA UNIV, SCH MED, DEPT NEUROL, MAEBASHI, GUMMA 371, JAPAN; TOHOKU UNIV, SCH MED, DEPT NEUROL, SENDAI, MIYAGI 980, JAPAN  
COUNTRY OF AUTHOR: JAPAN  
SOURCE: JOURNAL OF THE NEUROLOGICAL SCIENCES, (10 SEP 1997) Vol. 150, No. 2, pp. 133-135.  
Publisher: ELSEVIER SCIENCE BV, PO BOX 211, 1000

AE AMSTERDAM, NETHERLANDS.  
ISSN: 0022-510X.  
DOCUMENT TYPE: Article; Journal  
FILE SEGMENT: LIFE  
LANGUAGE: English  
REFERENCE COUNT: 8  
\*ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS\*  
AB McLeod syndrome is a rare X-linked disorder involving neurological defects and acanthocytosis. We examined the XK gene in three patients with neuroacanthocytosis, one of whom had cardiomyopathy, and his symptoms were very similar to those of McLeod syndrome. We found two new transversions (C to G at codon 204 and G to C at codon 205) in exon 3 in all those cases. However, the transversion at codon 205 was found in all 70 Japanese normal subjects and four non-Japanese (two Caucasian males, one Chinese female and one Micronesian female) and that at codon 204 was also detected in all 14 normal Japanese males and the four non-Japanese. These findings suggest that they are not the cause of McLeod syndrome, but normal polymorphisms which have not been reported. Moreover, there is a possibility that patients with neuroacanthocytosis similar to McLeod syndrome exist without the XK gene abnormalities. (C) 1997 Elsevier Science B.V.

L5 ANSWER 29 OF 36 MEDLINE DUPLICATE 13  
ACCESSION NUMBER: 94273191 MEDLINE  
DOCUMENT NUMBER: 94273191 PubMed ID: 8004674  
TITLE: Isolation of the gene for McLeod syndrome that encodes a novel membrane transport protein.  
AUTHOR: Ho M; Chelly J; Carter N; Danek A; Crocker P; Monaco A P  
CORPORATE SOURCE: Imperial Cancer Research Fund Laboratories, Institute of Molecular Medicine, John Radcliffe Hospital, Oxford, England.  
SOURCE: CELL, (1994 Jun 17) 77 (6) 869-80.  
Journal code: 0413066. ISSN: 0092-8674.  
PUB. COUNTRY: United States  
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
LANGUAGE: English  
FILE SEGMENT: Priority Journals  
OTHER SOURCE: GENBANK-Z32684  
ENTRY MONTH: 199407  
ENTRY DATE: Entered STN: 19940729  
Last Updated on STN: 20021022  
Entered Medline: 19940721

AB McLeod syndrome is an X-linked multisystem disorder characterized by abnormalities in the neuromuscular and hematopoietic systems. We have assembled a cosmid contig of 360 kb that encompasses the McLeod gene locus. A 50 kb deletion was detected by screening DNA from patients with radiolabeled whole cosmid, and two transcription units were identified within this deletion. The mRNA expression pattern of one of them, designated as XK, correlates closely to the McLeod phenotype. XK encodes a novel protein with structural characteristics of prokaryotic and eukaryotic membrane transport proteins. Nucleotide sequence analysis of XK from two unrelated McLeod patients has identified point mutations at conserved splice donor and acceptor sites. These findings provide direct evidence that XK is responsible for McLeod syndrome.

L5 ANSWER 30 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 14  
ACCESSION NUMBER: 1995:58381 BIOSIS  
DOCUMENT NUMBER: PREV199598072681  
TITLE: Efficiency of some serum protein systems in parentage control in Yugoslav trotter horses.  
AUTHOR(S): Trailovic, Ruzica; Jovanovic, S.; Savic, Mila  
CORPORATE SOURCE: Fac. Vet. Med., Univ. Belgrade, Bul. JNA 18, Belgrade Yugoslavia  
SOURCE: Acta Veterinaria (Belgrade), (1994) Vol. 44, No. 4, pp. 233-237.  
ISSN: 0567-8315.  
DOCUMENT TYPE: Article  
LANGUAGE: English  
SUMMARY LANGUAGE: English; Serbo-Croatian  
AB A total of 85 blood samples, obtained from Yugoslav trotter horses Were analysed for serum protein polymorphism at the following loci: albumin (Al), protease inhibitor (Pi), transferrin (Tf), esterase (Es) and \*\*\*Xk\*\*\* \*\*protein\*\*\* by standard starch gel electrophoretic procedures. From the results obtained the homogeneity index and parentage exclusion probability were calculated. The characteristic gene frequencies of the investigated Al, Pi, Ti, Es and \*\*\*Xk\*\*\* \*\*protein\*\*\* systems were established as: AIA and AIB (0.424 and 0.576); PiF, PiL, PiG, PiI, PiV and PiS (0.135, 0.318, 0.123, 0.100, 0.259 and 0.576); TiD, TiF, TiH and TiO (0.359, 0.529, 0.036 and 0.076), EsF, EsI and EsS (0.265, 0.570 and 0.165); and XKK and XKs (0.912 and 0.088), respectively. The Homogeneity index of the tested population was 0.0049, 0.5755, 0.2209, 0.1336 and 0.6790 for the AL, Pi, Tf, Es and Xk, loci, respectively. The joint paternity exclusion probability was 83.40% for the population of Yugoslav trotters.

L5 ANSWER 31 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 15  
ACCESSION NUMBER: 1993:294126 BIOSIS  
DOCUMENT NUMBER: PREV199396012351  
TITLE: A study on the polymorphism of blood protein and enzyme in Cheju native horses.  
AUTHOR(S): Kim, Sang-Yong; Kang, Min-Soo; Choung, Chang-Cho;  
Takahashi, Jutaro; Yasuda, Yasuhisa  
CORPORATE SOURCE: Fac. Agric. Iwate Univ., Morioka Japan  
SOURCE: Journal of the Faculty of Agriculture Iwate University, (1993) Vol. 21, No. 2, pp. 91-96.  
ISSN: 0579-2746.

DOCUMENT TYPE: Article  
LANGUAGE: Japanese  
SUMMARY LANGUAGE: Japanese; English  
AB On the basis of gene frequencies of the marker traits of blood protein and enzyme analyses with electrophoresis, the biochemical polymorphism of albumin (Al), slow-alpha-2 globulin (S1-alpha), post-albumin (Pa), group-specific component (Gc), \*\*\*Xk\*\*\* \*\*protein\*\*\* (Xk), transferrin (Tf), catalase (Cat), hemoglobin (Hb), phosphohexose isomerase (PHI), phosphogluconate dehydrogenase (PGD) and phosphoglucosutase (PGM), in a total 95 Cheju native horses, were examined. The analyzed results of phenotypes and gene frequencies were as follows: With respect to albumin (Al) locus, the frequency of Al-B allele was markedly predominant (0.663) as compared with that of Al-A allele (0.337). In slow alpha-2 globulin (S1-alpha-2) locus, any individual variation was not found. Therefore, this locus was defined to be monomorphic. In the post-albumin (Pa) locus, the frequency of Pa-F allele was markedly predominant (0.947) as compared with that of Pa-S allele (0.053). Concerning group-specific component (Gc)

locus, the frequency of Gc-S allele was markedly predominant (0.589) as compared with that of Gc-F allele (0.441). As to the \*\*\*Xk\*\*\* \*\*protein\*\*\* locus, one phenotype KK was observed. The number of the KK phenotype was 1.000. In the transferrin (Tf) locus, Tf-F was the most frequent allele gene frequency (0.621), Tf-R was the second (0.153) and Tf-H, Tf-D and Tf-O were negligible (0.131, 0.084, and 0.010). At the catalase (Cat) isozyme locus, the gene frequency of Cat-F allele (0.511) was slightly higher than that of Cat-S allele (0.489). In the hemoglobin (Hb) locus, the frequency of Hb-A allele (0.868) was remarkably higher than that of Hb-a allele (0.132). At the phosphohexose isomerase (PHI) isozyme locus, only phenotype II was observed. The frequency of the II type was 1.000. Phosphoglucumutase (PGM) isozyme locus, any individual variation was not found. As to phosphogluconate dehydrogenase (PGD) isozyme locus, any individual variation was not found.

L5 ANSWER 32 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.DUPLICATE 16  
 ACCESSION NUMBER: 1990:471454 BIOSIS  
 DOCUMENT NUMBER: BA90:110874  
 TITLE: STUDIES ON THE BIOCHEMICAL POLYMORPHISM OF BLOOD PROTEIN AND ENZYME IN CHE JU NATIVE HORSES I. GENETIC POLYMORPHISMS OF SERUM PROTEINS.  
 AUTHOR(S): CHUNG E Y; HAN S K; SHIN Y C; YANG K S  
 CORPORATE SOURCE: COLL. AGRIC., SANG JI UNIV., KOREAN.  
 SOURCE: KOREAN J ANIM SCI, (1990) 32 (6), 298-308.  
 CODEN: HGCHAG. ISSN: 0367-5807.  
 FILE SEGMENT: BA; OLD  
 LANGUAGE: Korean  
 AB By means of starch gel electrophoresis, the biochemical polymorphism of .alpha.1-protease inhibitor, albumin, transferrin, \*\*\*Xk\*\*\* \*\*protein\*\*\* and slow .alpha.2-globulin in a total of 116 Che Ju native horses were examined. The analyzed resulted of phenotype, genotype and gene frequency was following: 1. In the .alpha.1-protease inhibitor(Pi) locus, nine possible phenotypes, except heterozygous FI phenotype, FF, II, LL, SS, FL, FS, IL, IS and LS were identified and assumed to be controlled by four autosomal codominant alleles designated PiF, PiI, PiL and PiS. The phenotype distribution was estimated to be 68.10% for LL type and 12.93% for II type and the others were below 10%. The PiL allele with the frequency of 0.741 showed the highest frequency, while the frequencies of PiI, PiS and PiF alleles with relatively low frequencies were 0.164, 0.078 and 0.017, respectively. 2. With respect to albumin(AI) locus, three different AI phenotypes assumed to be controlled by two codominant alleles were identified as AA, AB and BB and their phenotype distribution was 15.52%, 40.52% and 43.96%, respectively. The frequency of AIB allele was markedly predominant (0.641) whereas in A1A allele it was 0.358. 3. Concerning transferrin(Tf) locus, eleven different phenotypes DD, FF, RR, DF, DO, DR, FH, FO, FR, HR and OR were recognized, assumed to be controlled by five autosomal codominant alleles designated TFD, TfF, TfH, TfO and TfR, but two homozygous type(HH and OO) and two heterozygous type(DH and HO) were not found. The observed percentage of Tf phenotypes FR, FF and RR were found to be 29.31%, 28.45% and 12.93%, respectively, and the other phenotypes were below 10%. Of the total, TfF was the most frequent allele(gene frequency, 0.496), TfR was the second(0.345) and TFD, TfO and TfH were negligible(0.065, 0.60 and 0.034, respectively). 4. As for the \*\*\*Xk\*\*\* \*\*protein\*\*\* locus, two different phenotypes FK and KK were observed, whereas homozygous FF type was not recognized. The observed Xk polymorphism was assumed to be controlled by a pair of codominant alleles designated XkF and XkK at a single autosomal locus. The number of the KK phenotype was 93.10, that of FK phenotype 6.90%. The significantly higher frequency of XkK allele(0.966) was obtained than that of XkF allele(0.034). 5. In slow .alpha.2-globulin(S .alpha.1) locus, any individual variation was not found, therefore, this locus was defined to be monomorphic.

L5 ANSWER 33 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.  
 ACCESSION NUMBER: 1990:284226 BIOSIS  
 DOCUMENT NUMBER: BA90:15072  
 TITLE: STUDIES ON BLOOD GROUPS IN RACE HORSES V. GENETIC POLYMORPHISM OF SERUM \*\*\*XK\*\*\* \*\*PROTEIN\*\*\* .

AUTHOR(S): HAN S K; CHUNG E Y; KANG H I  
 CORPORATE SOURCE: COLL. ANIMAL HUSBANDRY, KON-KUK UNIV., JPN.  
 SOURCE: KOREAN J ANIM SCI, (1990) 32 (2), 61-65.  
 CODEN: HGCHAG. ISSN: 0367-5807.  
 FILE SEGMENT: BA; OLD  
 LANGUAGE: Japanese  
 AB Genetic polymorphism of a new horse plasma protein provisionally designated \*\*\*Xk\*\*\* \*\*protein\*\*\* in 175 Korean race horses was analyzed by using acidic starch gel electrophoresis and genetic structure of horse population was investigated. Two different phenotypes, Xk-FK and Xk-KK. in this system were observed with the frequencies in these Xk phenotypes were Xk-FK 2.9% and Xk-KK 97.1%. However, the homozygous Xk-FF type was not recognized in the present study. Observed and expected phenotypes showed the Xk locus to be in genetic equilibrium, according to Hardy-Weinberg law. Therefore, the Xk phenotypes were shown to be controlled by two codominant autosomal alleles designated XkF and XkK. The XkK allele(0.986) had a remarkably high frequency whereas the XkF allele(0.014) occur very rarely.

L5 ANSWER 34 OF 36 MEDLINE DUPLICATE 17  
 ACCESSION NUMBER: 89250430 MEDLINE  
 DOCUMENT NUMBER: 89250430 PubMed ID: 3248368  
 TITLE: The homology between the serum proteins PO2 in pig, Xk in horse and alpha 1B-glycoprotein in human.  
 AUTHOR: Van de Weghe A; Coppieters W; Bauw G; Vandekerckhove J; Bouquet Y  
 CORPORATE SOURCE: Department of Animal Genetics, State University of Ghent, Merelbeke, Belgium.  
 SOURCE: COMPARATIVE BIOCHEMISTRY AND PHYSIOLOGY. B: COMPARATIVE BIOCHEMISTRY, (1988) 90 (4) 751-6.  
 Journal code: 2984730R. ISSN: 0305-0491.  
 PUB. COUNTRY: ENGLAND: United Kingdom  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 198906  
 ENTRY DATE: Entered STN: 19900306  
 Last Updated on STN: 19900306  
 Entered Medline: 19890628  
 AB 1. Pig serum Po2 protein and horse \*\*\*Xk\*\*\* \*\*protein\*\*\* were purified by FPLC, non-denaturing 2D agarose-PAGE and 2D IPG-PAGE. 2. The separated fractions were electroblotted to poly(4-vinyl-N-methylpyridinium iodide) coated GF/C glass fiber sheets. 3. The partial amino acid sequences and amino acid compositions of different genetic variants of the proteins were determined. 4. The results proved that previously reported polymorphic serum post-albumins in each of these species were homologous to human plasma alpha 1B-glycoprotein.

L5 ANSWER 35 OF 36 MEDLINE DUPLICATE 18  
 ACCESSION NUMBER: 83306728 MEDLINE  
 DOCUMENT NUMBER: 83306728 PubMed ID: 6614593  
 TITLE: Genetic linkage between the loci for phosphohexose isomerase (PHI) and a serum protein (Xk) in horses.  
 AUTHOR: Andersson L; Juneja R K; Sandberg K  
 SOURCE: ANIMAL BLOOD GROUPS AND BIOCHEMICAL GENETICS, (1983) 14 (1) 45-50.  
 Journal code: 0263344. ISSN: 0003-3480.  
 PUB. COUNTRY: Netherlands  
 DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)  
 LANGUAGE: English  
 FILE SEGMENT: Priority Journals  
 ENTRY MONTH: 198310  
 ENTRY DATE: Entered STN: 19900319  
 Last Updated on STN: 19980206  
 Entered Medline: 19831028  
 AB Genetic linkage between the equine loci for phosphohexose isomerase (PHI) and serum \*\*\*Xk\*\*\* \*\*protein\*\*\* was demonstrated by means of segregation data from three sire families. The recombination frequency was estimated from pooled data to be 0.23 +/- 0.02; a significant heterogeneity between sires for estimates of the recombination frequency was observed. No indication of linkage was detected between Xk and 14 other blood marker loci. Linkage between the Xk locus and the locus for soluble malic enzyme (ME1) has recently been reported in horses. An equine linkage group designated LG IV comprising the three loci ME1, PHI, and Xk has thus been established. The possibility that the linkage between PHI and Xk is homologous to the linkage between the loci for PHI and a serum postalbumin (PO-2) in pigs was discussed.

L5 ANSWER 36 OF 36 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC.  
 ACCESSION NUMBER: 1984:185837 BIOSIS  
 DOCUMENT NUMBER: BA77:18821  
 TITLE: EQUINE GENE MAPPING CLOSE LINKAGE BETWEEN THE LOCI FOR SOLUBLE MALIC ENZYME EC-1.1.1.40 AND XK PA.  
 AUTHOR(S): WEITKAMP L R; COSTELLO-LEARY P; GUTORMSEN S A  
 CORPORATE SOURCE: DEP. PSYCHIATRY, DIV. GENETICS, UNIV. ROCHESTER SCH. MED. DENT., 601 ELMWOOD AVE., ROCHESTER, N.Y. 14642, U.S.A.  
 SOURCE: ANIM BLOOD GROUPS BIOCHEM GENET, (1982 (RECD 1983)) 13 (4), 279-284.  
 CODEN: ABBGBX. ISSN: 0003-3480.  
 FILE SEGMENT: BA; OLD  
 LANGUAGE: English  
 AB Resolution of equine soluble malic enzyme phenotypes is greatly improved by isoelectric focusing as compared with starch gel electrophoresis. Phenotype differences can be recognized in plasma as well as hemolysates. The locus for soluble malic enzyme (ME1) is closely linked to the locus for \*\*\*Xk\*\*\* [ \*\*protein\*\*\* ].